

The development of language and
number understanding in
Williams Syndrome and Down's
Syndrome:

Evidence from the infant and mature
phenotypes

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Abstract

This thesis is an examination of language and number in two atypically developing groups, Williams Syndrome (WS) and Down's Syndrome (DS). These groups were chosen because their cognitive profiles in adulthood differ significantly. It is already known that language is a relative strength in WS but that it is poorer than non-verbal ability in DS.

The precursors to both language and number ability were studied in 24-36 month old infants and performance at this stage was compared with that in the steady state, by testing older children and adults, aged 9-35 years. Similar age-appropriate tests were used with both groups so that performance in the steady state could be compared with that in infancy. Specific subdomains of language and number were assessed to investigate whether the pattern seen in the adult steady state was also present in infancy, or whether the mature phenotype is a product of the different developmental trajectories followed by each group.

The overall cognitive profile of infants with WS and DS did not differ significantly, despite clear distinctions between the adult profiles. However, their performance on number and language tasks did differ in infancy. While in adulthood WS performance on number tasks was poorer than that of DS, in infancy this pattern was reversed. For language, infants with DS exhibited a large discrepancy between productive and receptive vocabulary. A more even pattern was present for the WS group. In adulthood, vocabulary was better in WS than DS but both groups had problems with syntactic structures. Taken together these results suggest that it is not possible to derive the pattern of infant performance from the steady state in adulthood. The developmental trajectories from precursors to mature phenotype need to be thoroughly charted in atypical populations because the study of development, not just the characterisation of the endstates, is crucial.

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Introduction

The cognitive phenotypes of neurodevelopmental disorders, such as Williams Syndrome and Down's Syndrome, have hitherto been characterised in a snap shot fashion at various points in development. However, very few studies have attempted to chart their developmental trajectories. The research in my thesis aims to begin to take a truly developmental approach by examining the infant cognitive abilities of these two atypically developing groups: one that is well-known and highly researched, Down's Syndrome (DS), and the other that is much rarer, Williams Syndrome (WS). In other words, instead of merely characterising abilities in the steady state when the phenotype has reached maturity, my research attempts to make links between this state and the nature of specific abilities at the outset of development, in infancy. Only when the infant state is fully characterised, can we begin to understand the effects of development on disorders such as DS and WS. Ultimately, it will be necessary to chart the full developmental trajectories of these disorders to ascertain how they differ from each other and from the typical pattern.

1.1 Populations studied

The aim of this research is to investigate the language and numerical abilities in infancy and in adulthood of two atypical groups. The syndromes were chosen because they exhibit distinctive cognitive phenotypes in later life. Williams Syndrome has a very uneven cognitive profile with marked strengths and weaknesses. Similarly, although the profile is less extreme, peaks and troughs of ability are present for Down's Syndrome. Before the specific issues to be tackled in this thesis are introduced, the following sections will describe the genotype, brain structure and cognitive phenotype of Down's Syndrome and Williams Syndrome.

1.1.1 Down's Syndrome

Down's Syndrome (DS) is a genetic disorder, occurring in approximately 1 in 800 live births. This incidence increases with maternal age to approximately 1 in 600 births (Dolk et al., 1990). The disorder gives rise to clear physical characteristics and learning disabilities, with IQs in a similar range to those found in WS.

Physical characteristics of Down's Syndrome

Along with facial dysmorphology come other physical problems, such as congenital heart disease, respiratory tract infections, poor hearing and vision (Courage et al., 1994; Marcell & Cohen, 1992), lack of muscle tone and motor problems (Henderson, 1985), and thyroid disease. Individuals with DS have smaller heads which are flatter at the back than those found in the general population. They have upwardly slanting eyes with marked epicanthic folds and short necks.

Genetics of Down's Syndrome

Down's Syndrome results from defects on the 21st chromosome, the most common of which is trisomy 21 (47 XX or 47 XY) in which an extra chromosome 21 is present (Lejeune et al., 1959). This is found in approximately 95% of people with Down's Syndrome. Translocations are found in 1-5 % of cases and usually appear between chromosomes 14 and 21. Mosaic forms are found in about 1-2 %, with some cells having trisomy 21 and others not.

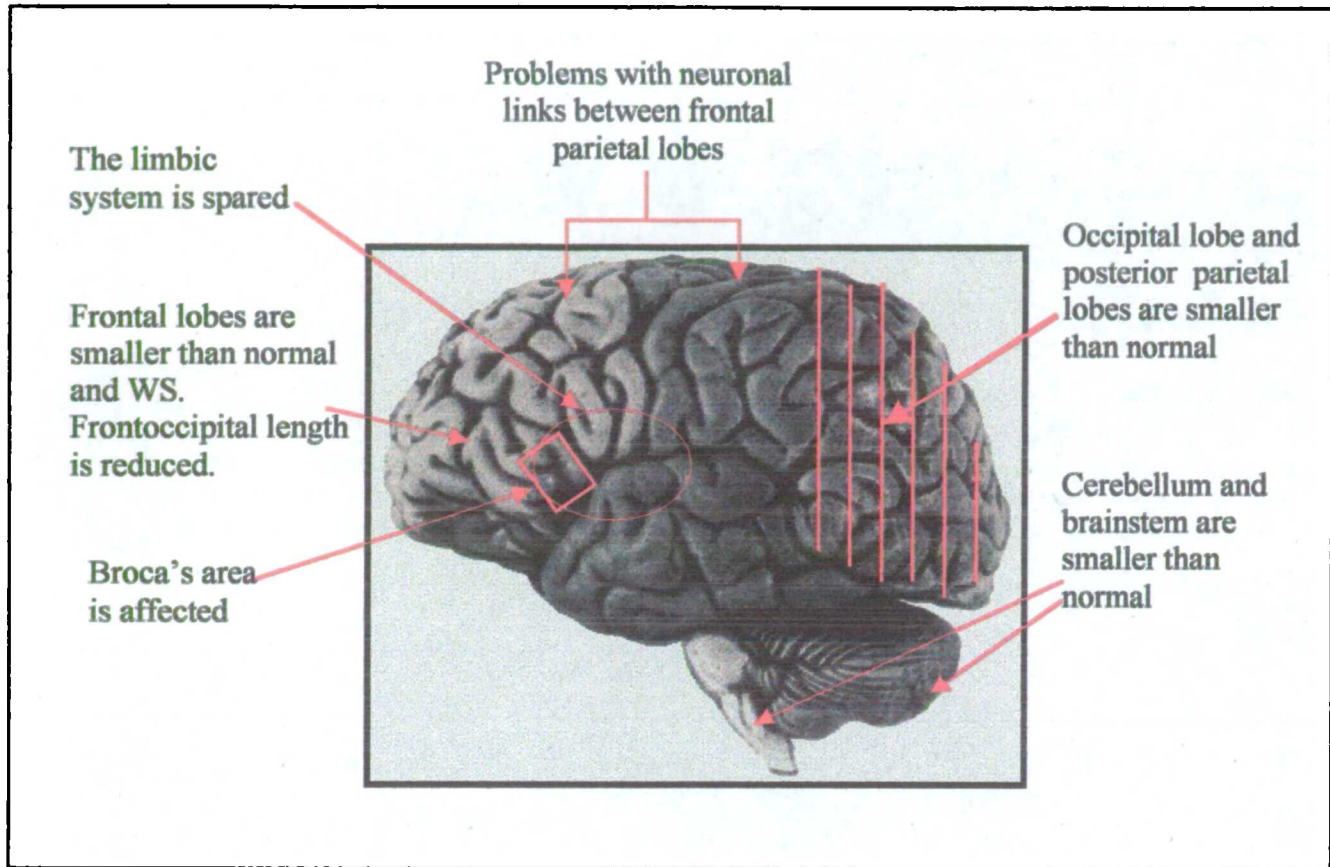
The individuals who took part in the research reported in this thesis all have Down's Syndrome of the trisomy 21 type. They were chosen because the other types of Down's Syndrome give rise to a much more varied phenotype. In an already highly variable population, it was decided that the variation should be controlled as much as possible. In addition, there is a greater incidence of trisomy 21 DS than for the other two types, and it was therefore easier to recruit participants with trisomy 21.

Brain morphology in Down's Syndrome

Chromosomal abnormalities have an effect on the DS brain, which develops abnormally, see Figure 1.1. Unlike for WS (which will be described later), there are data on the structure and volume of both DS infant and DS adult brains, from both autopsy and functional imaging studies. These data suggest that very early in development, prenatally and up to a few months postnatally, there is little structural difference between a DS and a typically developing brain (Brooksbank et al., 1989; Florez, 1990). However, studies have indicated that in 25% of infants with DS there is a delay in the myelination of those nerve fibres which are myelinated late in development, between 2 months and 6 years of age (Wisniewski, 1991). This particularly affects neurons in the frontal and temporal lobes. In addition, by 3-5 months after birth, the frontal lobes of the brain are proportionally reduced in volume and, in most cases, the brain stem and cerebellum appear small. There also seem to be abnormalities in the auditory system, with delayed arborization of neurons in this area (Jiang et al., 1990), suggesting that neural connections may be impaired. In contrast, there is a larger amount of dendritic arborization in the visual system of a DS infant compared to normal infants, but by 2 years of age there is less than normal (Backer et al., 1986).

In later childhood and adulthood, the volume of mesial temporal lobe structures is particularly reduced (Jernigan, Bellugi, Sowell et al., 1993). And there are abnormalities in the link between frontal and parietal areas, which may include Broca's area, an area implicated in typical language function (Schapiro et al., 1992). Studies of metabolic rate using PET indicate that there is a higher cortical metabolic rate in DS adults (Haier et al., 1995). It is also known that the DS brain deteriorates with age, with 100% of older adults with DS exhibiting signs of neuropathology like those seen in Alzheimer's disease, though only about 50% actually develop the symptoms (Nadel, 1990). The atypical brain morphology may provide some basis for the resulting DS cognitive profile.

Figure 1.1 The Down's Syndrome Brain



Cognitive phenotype in Down's Syndrome

Cognitive development in DS is generally slower than normal, with MA at about one half to two-thirds the normal level and reaching a plateau at the level of a typically developing 6-8 year old (Carr, 1985; Wishart & Duffy, 1990). DS was chosen as a comparison group for WS because these groups exhibit a similar overall level of mental retardation and undergo similar early intervention programs (Bellugi et al., 1996). Like WS, DS gives rise to an uneven cognitive profile, although the unevenness is less marked than in the WS profile. What makes the comparison interesting is that the peaks and troughs in the DS profile differ from those in the WS profile, and are in some respects the opposite.

The most striking feature of cognitive development in DS is that language ability lags behind other cognitive abilities (Fowler, 1990; Rosenberg & Abbeduto, 1993). These

differences are apparent by 5 years. They are most obvious in productive language, but also occur in receptive language (Lynch & Eilers, 1991; Miller 1992a, 1992b.) Individuals with DS have particular problems with grammatical development but also have smaller vocabularies than normal. Some of the language difficulties may be exacerbated by auditory problems, such as chronic otitis media, varying degrees of deafness (Marcell et al., 1992, 1995), and problems with poor articulation (Dodd, 1997). In contrast to WS, individuals with DS also have impaired phonological short term-memory (Jarrold, Baddeley & Hewes, 1998).

In comparison with WS, strengths in the DS cognitive profile include visual short-term memory (Jarrold et al., 1998) and performance on visuo-spatial tasks such as picture copying. Individuals with DS produce pictures which retain the overall configuration of elements but produce less featural detail, the converse of the WS pattern (Bihrlé et al., 1989).

There are also elements of cognitive functioning which are at the same level in both WS and DS. Both groups have difficulty with number (Klein & Mervis, 1999) and Piagetian conservation tasks (Bellugi, Bihrlé, Neville, Doherty & Jernigan, 1992). Although language is a relative strength in WS, both groups experience language and motor delays. Attentional difficulties are also common to both groups (Fisher, 1970; Nesbitt, Howlin & Udwin, 1999), and must be taken into consideration when devising empirical studies of cognitive function.

Now that the main characteristics of DS have been introduced, I turn to Williams Syndrome.

1.1.2 Williams Syndrome

Williams Syndrome (WS) is a rare contiguous gene disorder found in approximately 1:20000 live births. It is caused by sub-microscopic deletions on the long arm of chromosome 7q11.23. These deletions include the elastin gene missing in 98% of those with WS (Ewart, Morris, Atkinson, Jim, Sternes, Spallone et al., 1993) and the

Lim-Kinase 1 gene (Frangiskakis, Ewart, Mervis, Bertrand, Robinson et al., 1996; Tassabehji, Metcalfe, Fergusson, Carett, Dore, Donnai et al., 1996).

Physical characteristics of Williams Syndrome

The physical characteristics of WS include facial dysmorphology and cardiovascular defects. Individuals have "elfin facies", characterised by a broad brow, wide mouth, full cheeks, stellate irises, dental irregularities and a low nasal root (Beuren, 1972; Morris et al., 1988; Williams et al., 1961). Other physical problems include connective tissue abnormalities, caused by the deletion of the elastin gene. These abnormalities may give rise to cardiac and vascular defects such as pulmonary aortic stenosis and supralvalvar aortic stenosis (SVAS). In addition, there is a high incidence of inguinal hernias in this population. Higher than normal levels of blood serotonin have also been reported (August & Realmuto, 1989; Marriage, 1995) which might be linked to neurological abnormalities.

Infants with WS often have problems with feeding, including hypercalcaemia, which result in failure to thrive during the early months of development. In later life, people with WS are of short stature and can experience joint problems (Udwin & Yule, 1991). Up to 90% of individuals with WS have hyperacusis, or extreme sensitivity to sound, which can lead to problems in everyday activity (Marriage, 1995). This occurs despite a lack of obvious organic cause. As mentioned in the later section on the WS brain, there are suggestions that Heschel's gyrus is abnormally large in people with this syndrome which could explain the hyperacusis.

As with all phenotypes, the characteristics exhibited in different individuals vary. However, this variation is unlikely to depend on the size of the underlying deletion, because its size is identical in 90% of cases. Instead, the individual differences must be due to the familial genetic potential of the individual and the way in which the environment has affected the genotype. The genotype is described in the next section.

Genetics of Williams Syndrome

WS is caused by a hemizygous microdeletion of about 1.5 Mb on chromosome 7q11.23. As mentioned above, the size and position of the deletion is very similar across individuals because the area containing the genes deleted in WS is flanked by areas in which genes are repeated (Baumer et al., 1993). During recombination, these repeat sequences become misaligned on one allele and the area between two repeated sequences is spliced out and therefore lost.

Several genes in the deleted area have now been characterised. The Elastin gene encodes the protein elastin, and has been implicated in the connective tissue impairments in WS (Ewart et al., 1993). Individuals without WS but with an elastin deletion have SVAS, supralvalvular aortic stenosis; therefore, ELN can be linked to the cardiological problems exhibited in WS. However, despite its function, the elastin gene does not appear to cause the facial dysmorphology because the SVAS patients do not exhibit atypical facial characteristics of WS (Tassabehji et al., 1996; 1999).

LIMK1, the lim-kinase 1 gene, is also deleted in WS (Tassabehji et al., 1996). This gene, LIMK1, encodes for a protein used in brain development and large amounts are found in the CNS (Okano et al., 1995). Deletions of genes expressed in the brain and CNS, such as LIMK1, could be at the root of cognitive impairment, so these genes are ideal candidates for study across conditions which share part of the WS deletion. The deletion of LIMK1 in SVAS provides a tool for investigating which genes might be implicated in the overall cognitive profile found in WS. If LIMK1 were a candidate for the cause of cognitive impairment then the same characteristics would be shared by individuals with WS and those only with SVAS. This was not the case for the four patients with SVAS studied by Tassabehji et al. (1996), three of whom performed in the normal range on a standardised ability test and did not exhibit the characteristic Williams Syndrome cognitive profile (WSCP), i.e. poor spatial cognition and relatively good language. Instead, they performed equally well on both spatial and language tasks. This suggests that LIMK1 is not a necessary and sufficient candidate gene for cognitive impairment of the WSCP. By contrast, previous work by Frangiskakis et al. (1996) provided data, which suggested that individuals from two

families with both ELN and LIMK1 deletions did exhibit the WSCP. However, it should be noted that these data come from only two families and it may be that the families are examples of two specific deletions and not the effects of LIMK1 per se. The later results from Tassabehji and colleagues suggest that the conclusions of Frangiskakis et al. cannot be generalised.

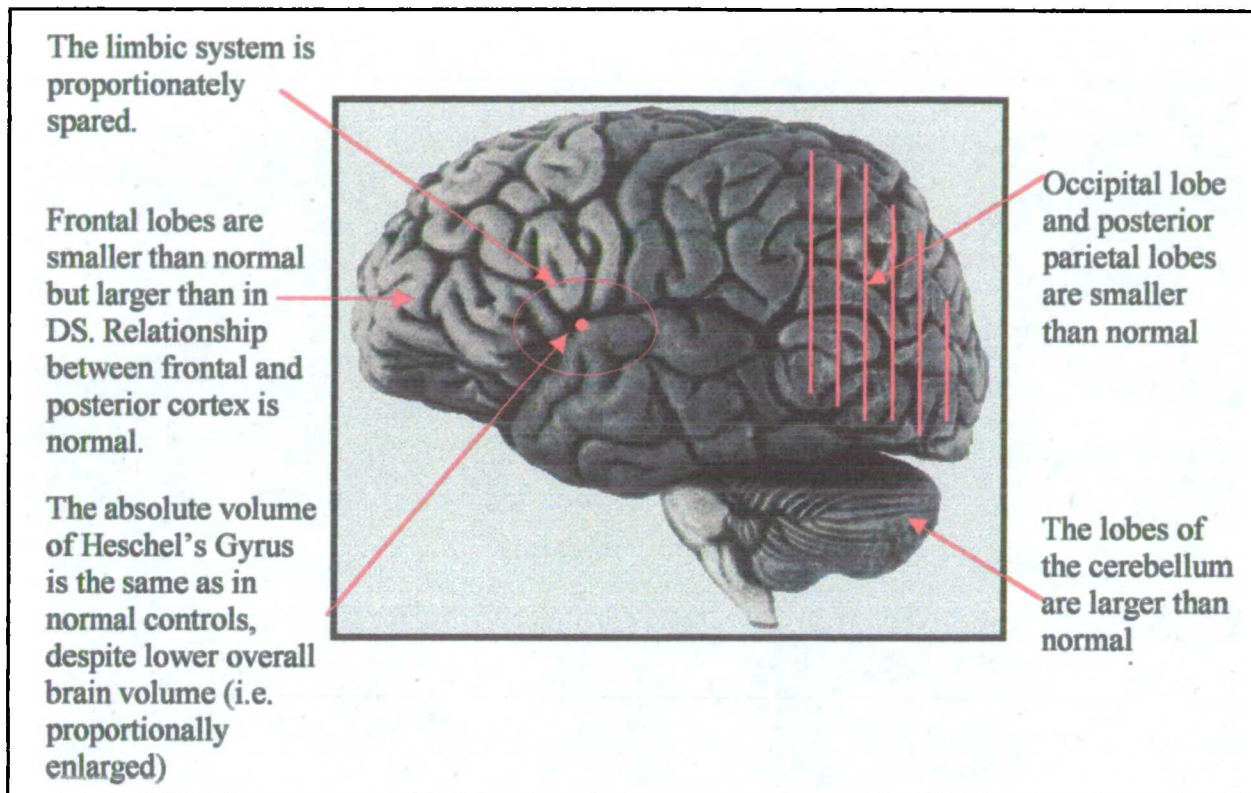
In addition to ELN and LIMK1 the following genes have been identified in the critical region for WS: syntaxin (STX1A) which encodes part of the synaptic mechanism (Osborne, 1997), Frizzled (FZD3) gene (Wang et al., 1997), RFC2 - replication factor C2 (Peoples et al., 1996) and WSCR1, 8 and 9, WS BHLH, CPETR 1 and 2, CYLN2, FKBP6, BCL7B, TBL2, GTF2I and NCF1. The role of these genes is not yet known and several more genes in the deleted region are yet to be identified. Even when the genes in the deletion are characterised, it is important to remember that there are unlikely to be simple genes to behaviour mappings. Gene expression is very complex, and genes interact with one another and with their environment, so their relationship to the development of certain cognitive processes and behaviours is very indirect.

Although in the past, diagnosis of WS was only carried out clinically and many cases were misdiagnosed, WS can now be detected in early infancy. The FISH test (fluorescent in-situ hybridisation) is used to probe for the deletion of the Elastin gene, which is a core deletion in WS. All participants in this thesis were positive on the FISH and all had a confirmed clinical diagnosis.

Brain morphology in Williams Syndrome

The deletions that cause WS have an effect on many aspects of the individual including brain development. The WS brain is about 80% of that of a typical brain and consequently there is a lower volume of cortical grey matter. There are also some differences in proportions of different brain structures, which are illustrated below in Figure 1.2.

Figure 1.2 The Williams Syndrome Brain



As illustrated, the ratio of frontal to posterior cortex volume is near normal although both are reduced (Jernigan, Bellugi, Sowell, Doherty & Hesselink, 1993; Wang, Hesselink, Jernigan, Doherty & Bellugi, 1992). Caution should be exercised when inferring WS function from the normal case, because WS brains are likely to develop differently from the brains of normal controls. However, data from participants with WS support the hypothesis that there may be a link between the volume of the frontal lobes and the level of language ability of an individual (Leiner et al., 1993). Those individuals with the largest volume of inferior frontal cortex, as measured by structural MRI, also performed better on standardised language tests (Jones et al., 1995). The volume of Heschel's gyrus, an area important for auditory processing in the typically developing case, is not reduced in WS. In fact, its absolute volume is the same as in normal controls (Hickock et al., 1995) and thus its relationship to the reduced volumes of the rest of the WS brain is very abnormal. Although no direct investigations have been undertaken, as mentioned earlier, this enlargement of

Heschel's gyrus might be linked to the atypical auditory processing in WS (Neville, Mills & Bellugi, 1994). Data from studies measuring Event Related Potentials to auditory stimuli provide supporting evidence for this suggestion. Individuals with WS produce abnormally large positive electrophysiological responses (P50 and P200) over temporal areas of the brain when presented with auditory stimuli. These responses are not found in the normal population, in either childhood or adulthood (Neville et al., 1994).

Support for a cautious approach to linking brain structures directly to behaviour in atypical populations is provided by neurochemical data from an investigation of the cerebellum by Rae, Karmiloff-Smith, Lee, Dixon et al. (1998). Despite the proportionally normal volume of the cerebellum in WS, brain chemistry in this area is abnormal.

In addition to volumetric and neurochemical abnormalities described above, there are data that indicate differences in the cytoarchitecture of WS brains. These cell data come from a small sample of autopsies, so further research needs to be conducted before generalising to the whole WS population. Galaburda et al. (1994, 1995) found a low density of cells in the cortex and, where there were fewer neurons, there was a larger number of glial cells. This decrease in cell packing density suggests that either brain development may have been curtailed abnormally early, or that the brains may have already begun to degenerate in the patients from whom these autopsy samples were taken.

The organisation of cells in the visual cortex is particularly abnormal. Instead of following the usual pattern of vertical layers, the neurons in WS visual cortex follow a more horizontal pattern (Galaburda, Wang, Bellugi & Rossen, 1994). This might be at the root of the visuo-spatial problems encountered by individuals with WS. The cognitive phenotype which results from these brain abnormalities is described in the next section.

Cognitive phenotype in Williams Syndrome

Williams Syndrome results in an unusual cognitive and personality profile. Individuals generally have mild to moderate learning difficulties with IQs in the 50s and 60s (ranging from 40-90). They are generally unable to live fully independently (Nesbitt, Howlin & Udwin, 1999). They are particularly interested in people, very sociable and empathic (Karmiloff-Smith et al., 1995; Tager-Flusberg & Sullivan, 1996) but can have problems with anxiety (Udwin & Yule, 1991). However, it is their uneven pattern of cognitive strengths and weaknesses that is striking and of particular interest here (Bellugi, Lichtenberger, Jones, Lai, St. George, 2000; Karmiloff-Smith, 1998; Karmiloff-Smith, Grant, Berthoud, Davies, Howlin & Udwin, 1997; Klein & Mervis, 1999; Mervis, 1999; Tager-Flusberg & Sullivan, 1996; Vicari, Carlesimo, Brizzolara, & Pezzini, 1996; Volterra, Capirci, Pezzini, Sabbadini & Vicari, 1996). WS has been singled out by many theorists (e.g., Pinker, 1991; 1994) because of this uneven profile, in which some cognitive abilities, such as language, appear “intact” while others, such as visuospatial construction are “impaired”. WS is used as support for a modular basis of cognition, in which certain aspects of cognition, or modules, are argued to be spared in the face of a disorder where others are affected.

It is true that relative to overall mental age level, some aspects of WS cognition are better or worse than others. Those abilities that are relatively good in comparison with what would be expected given mental age level include: some aspects of language, such as vocabulary (Karmiloff-Smith et al., 1997), phonological short-term memory, such as digit-span (Mervis et al., 1997), and face recognition (Karmiloff-Smith et al., 1997; Udwin & Yule, 1991). However, even within so-called spared abilities, there are within-domain strengths and weaknesses. Within the domain of language, for example, vocabulary is a relative strength, whereas there are problems with syntax and morphology (Karmiloff-Smith, Grant, Berthoud, Davis, Howlin & Udwin, 1997; Karmiloff-Smith et al., 1998). In addition, although vocabulary is relatively good, there is evidence to suggest that adults with WS do not employ all of the normal constraints when learning new words (Stevens & Karmiloff-Smith, 1997).

Those areas of cognition in which performance is particularly weak relative to mental age level include: visuo-spatial construction, as measured by tasks such as block design (Bellugi et al., 1990), planning and executive function, number skills (Bellugi et al., 1988, 1992, 1994; Karmiloff-Smith, 1992; Karmiloff-Smith, Klima, Bellugi, Grant & Baron-Cohen, 1995, Klein & Mervis, 1999, Paterson et al., 1999) and visuo-spatial memory (Vicari, Brizzolara, Carlesimo, Pezzini, and Volterra, 1996).

Impairments on visuo-spatial constructive tasks are likely to stem from the cognitive processes that are part of the WS phenotype. Individuals with WS appear to focus on the local properties of spatial arrays, so for example their drawings may depict the necessary components of an object but not in the correct configuration (Bihrlé et al., 1989). Interestingly, while this cognitive style causes impaired behaviour on construction tasks, for face processing it gives rise to good performance, but by an atypical means. Adults with WS do well on the Benton Facial Recognition Test (Benton, Hamsher, Varney & Spreen, 1983) but they appear to use componential, rather than configural processes to arrive at their response (Karmiloff-Smith, 1997; Udwin & Yule, 1991).

The fact that individuals with WS do well on face recognition tasks does not necessarily mean that there is a face recognition module which is intact, as some have claimed (Rossen, Klima, Bellugi, Bihrlé & Jones, 1995). The link between cognitive processes and behaviour is more subtle. While these individuals reach “normal” behavioural scores on these tasks, the processes underlying this behaviour are atypical and can contribute to impairments in other areas. This highlights the importance of making a rigorous investigation of atypical cognitive development. It is not sufficient to look at endstates, either over the life span or, more subtly, when looking at individual aspects of behaviour.

This pattern of relative strengths and weaknesses in childhood and in adults with WS and DS makes it crucial to investigate their precursors in infancy. (The term precursor is discussed in section 1.2.4.) Do areas of competency develop differently from areas of weakness, and are these differences present from the outset or do they emerge as a product of development? These questions are central to the aims of this thesis.

1.2 Methodological issues

1.2.1 Control Groups

In addition to the two clinical groups, two control groups were formed for both the infant and adult populations: a mental-age-matched group (MA-matched) and a chronological-age-matched (CA-matched) group. Atypical infants formed a pair, with a Down's Syndrome and Williams Syndrome infant matched on chronological age, mental age, sex, and as far as possible socio-economic status. These pairs were matched to both a MA- matched typically developing infant and a CA-matched typically-developing infant. This was done for a number of reasons. The MA-matched group was chosen because it would give an indication of typical overall performance at a given developmental level. This enables one to ascertain whether performance in the DS or WS groups is due to the overall cognitive impairment or to syndrome-specific differences in cognition. The inclusion of an MA-matched group is especially important for new tasks for which there are no previous details of typical performance. However, this approach does not take amount of experience into account as the MA-matched infants are usually about half to two-thirds below the chronological age of the atypically developing infants. In order to address this, a group of CA-matched infants was included. These infants were assumed to have the same amount of experience as the atypical groups, but were of a higher developmental level.

There are a number of difficulties when ascertaining a match for atypically developing individuals. This is exacerbated when the cognitive profiles of the groups are uneven, as is the case for WS and DS. There is much debate concerning which measures should be used for matching (Mervis & Robinson, 1999). If a measure with a large language component is used, in order to get an overall match between DS and WS, the DS group would have to be of a higher overall developmental level than the WS group, in order to compensate for the DS language disadvantage. By contrast, if matches are made on tests with a large bias towards spatial skills, then the WS group would have to be at an overall higher level. For this thesis, mental age was obtained from a widely used, standardised ability test, the Mental Scale of the Bayley Scales of

Infant Development II (Bayley, 1993). This test encompasses a wide range of cognitive abilities and, while it is not without problems (see chapter 2), it is the most commonly used test in the field of infant development. It was useful for the present investigation because it allowed performance to be compared across the domains of language, non-verbal ability, social, and motor skills.

Adults were matched on overall mental age, taken from the British Ability Scales (Elliott, Smith, McCulloch, 1996). This test was chosen because it is made up of several subtests which can be used to assess the strengths and weaknesses in the cognitive profile of individuals. In addition, the American version of this test, the Differential Ability Scales, has been employed in studies by Carolyn Mervis and colleagues (1999), which I hoped to replicate. As in the infant studies, participants were matched to a group of typically developing individuals on chronological age and to another group on mental age.

1.2.2 Age groups examined

The research examined number and language ability at two stages. The first and most important was the infant starting state, in which precursors were examined. The steady state, in middle childhood and adulthood, where the product of development can be examined, was also investigated. The main focus of this thesis is the infant group. This is because much less is known about cognitive ability at the outset of development in DS and WS than in the phenotypic outcome. The characterisation of the initial state is crucial for the study of atypical development. This enables one to investigate whether there is a point at which development deviates from the normal pathway or whether developmental processes are different from the very outset. If one examines only the steady state, when the mature phenotype is reached, then it is impossible to ascertain the role of development itself in contributing to the formation of the cognitive phenotypic outcome.

Ideally, studies of the infant phenotype should start at birth or in the early months of life. However, it was not possible to test very young infants because of the health problems encountered by infants with WS in the early months of life. Therefore,

infants were seen at 24, 30 and 36 months. Some infants were seen on two or three occasions, while others were tested only once. These ages were chosen so that it would be possible to chart development over the course of a year, as early as feasible in atypical development.

A group of older children and adults was included because the mature cognitive phenotype has not been fully explored in some domains. Little is known about the number skills of adults with Williams Syndrome, so it was necessary to make a preliminary investigation of them. In addition, older subjects were included so that the findings in the literature could be verified using a British sample. Much of the previous research into WS has been carried out in the USA, using American versions of standardised tests. Before arguments could be made pertaining to the contrast between adult and infant profiles of development in atypical groups, it was necessary to clarify the cognitive characteristics of the mature phenotype in these groups. This group of children and adults spanned a wider range than would be ideal, but given the rarity of WS, it was necessary to include all suitable subjects in order to achieve an acceptable sample size.

1.2.3 Testing clinical groups

In addition to the problems of matching and control groups, there are problems with testing and recruiting individuals with Down's Syndrome and Williams Syndrome. As mentioned in the section on WS, this syndrome is very rare and so it can be difficult to create a large sample. This is particularly true for the infant group. Until recently, it was unusual for individuals with WS to be diagnosed in infancy, so infant research was not possible. Now, diagnosis using the FISH test can be made in infancy but problems with infant recruitment nonetheless remain. Many infants with WS have feeding problems and experience failure to thrive. Consequently, the sample in this thesis had to be beyond their first year. At 24-36 months, the children had recovered from earlier physical problems. Despite the infants not being as young as ideal, this is the first research to assess infants with WS using a wide variety of tests.

Individuals with WS and DS also have sensory deficits and these must be taken into account when recruiting participants. However, attentional problems are the main obstacle when considering tests for use with these groups. Test administration takes much longer with atypically developing than control groups (see Chapter 2) because it is much more difficult to keep such individuals on task. This means that test sessions have to be planned accordingly and participants seen on a number of occasions.

Tests also have to be devised and adapted to the participants' chronological age. Although the participants with WS and DS had mental ages well below their chronological ages, and so were functioning like infants as young as 11 months, the atypical toddlers did not behave like infants. While a typical 11-month old infant will sit relatively passively and watch visual displays, a 24-month-old toddler with WS will be much more active and restless. This behaviour presents a challenge when using looking paradigms which are MA but not necessarily CA appropriate. The demands of atypically developing infants mean that testing these groups is both more time consuming and more demanding on research helpers than is the case for normally developing individuals. Specific issues relating to individual tasks will be discussed in the appropriate chapters of this thesis.

Irrespective of syndrome, there are problems with testing infants, which include participant drop-out due to excessive fussing and difficulties in keeping the participant on task.

1.2.4 Precursors

It is important to discuss what is meant by the term precursor. In the context of comparing adult cognitive profiles to infant profiles, a precursor can be thought of as an indicator or marker of the same pattern in adulthood.

In discussions concerning particular tasks or domains this meaning is slightly different. A precursor of an adult ability does not necessarily entail the same cognitive processes as its mature counterpart. Rather, it is likely to be the foundation or emerging ability from which the mature process or representation stems. This means

that the infant processes used in a particular situation may not be the same as those seen in the steady state. This is because the representations can alter as development progresses (Karmiloff-Smith, 1992b). For example, in the chapter on infant number, the infant task (making a discrimination between a novel and familiar numerosity) could be thought of as a precursor to and as sharing some characteristics with the adult task, which involves choosing the larger of two numbers. However, the cognitive processes used in each case are not necessarily equivalent. The infant could be employing domain general strategies which are number-relevant, whereas the adult is likely to be relying on processes relating directly to number and its meaning. The infant and adult mechanisms may be two instances of processing number-relevant stimuli but at two different points in developmental time. These changes over time may be particularly important when considering atypical development.

1.3 Theoretical Issues

Having presented a brief description of each syndrome, the control groups, and methodological problems, let us now turn to the main theoretical issues considered in the thesis.

As mentioned briefly in the introductory paragraph, much of the research into atypical cognitive function has concentrated on characterising the mature phenotype. While this is important, this approach tells us very little about the processes which give rise to the pattern of abilities present in these groups. DS and WS were chosen for this thesis because they differ on some critical aspects of cognition and are at a similar level of development on others.

As previously mentioned, WS, with its uneven pattern of strengths and weaknesses, has been used by some theorists to support claims about innate and independently functioning modules (e.g., Pinker, 1991; 1994). Several researchers believe that the static neuropsychological model derived from data from adults with brain damage can be applied to developmental disorders (Baron-Cohen, 1998; Leslie, 1992; Temple, 1997). Their argumentation implies that patterns of strengths and weaknesses in the mature phenotype are necessarily present in the initial state. For example, WS is seen

as having an intact language module, an intact face processing module and an impaired spatial cognition module, with relative strengths seen as intact rather than merely less impaired (Pinker, 1991; 1994). Given the pattern reported for WS and DS, this view would predict that infants with DS would have impaired language and number and that infants with WS would have intact language but impaired number. However, this approach based on damage to a previously normal adult brain is not appropriate for neurodevelopmental disorders.

In contrast to the static approach to development outlined above, this thesis will examine the hypothesis that individuals with WS and DS develop differently from the very outset. It is inappropriate to think of their impairments in terms of missing or intact modules, but instead they should be thought of as the product of different developmental trajectories (Elman et al., 1996; Karmiloff-Smith, 1998; Karmiloff-Smith, Brown, Grice & Paterson, in press; Paterson et al., 1999). Differences in genes on one or both alleles, whether reductions or increases, have indirect and subtle effects on the function of other genes and on the way in which the developing organism interacts with the environment. A dynamic neuroconstructivist approach to development suggests that the developing organism does not have pre-specified modules but instead has mechanisms which are more suited to particular types of inputs and processing than others. These biases in mechanisms lead to their increasing specialisation as a product of development rather than as its starting point (Elman et al., 1996; Karmiloff-Smith, 1992). By this view, it is unlikely that impairments or enhancement of behaviour can be linked directly to specific genes. For example, very small differences in the timing of events in brain development, such as the start of neural migration, could lead to very large differences in cognitive function. More generally, in atypical development, genetic abnormalities will lead to subtle changes in the developmental trajectory of the individual and this in turn will alter the processes which bring about the gradual specialisation of cognitive function. It might be that there is over-specialisation for some functions, which lead to strengths in some areas and impairments in others, or that there is less specialisation which could also lead to impairments (Karmiloff-Smith, 1998). For example, the brain of an infant with WS might come to the world with a preponderance of neural structures that are particularly suitable for the processing of linguistic input or these mechanisms might

be particularly efficient in WS. This may cause the infant to seek out linguistic stimuli to a greater extent than other forms of input and may lead to the fine tuning of large amounts of the processing resources to language-like input. Because large amounts of the processing capacity of such a brain might be given over to linguistic information, a strategy of storing information on an instance-by-instance basis would be viable. However, such a strategy would mean that the amount of rule abstraction necessary would be reduced and in turn such a processing style might become less efficient.

Given that small differences in the early stages of development can cause infants to follow radically different developmental trajectories, is it possible to infer the infant cognitive profile from the profile in the adult endstate? This would be a consequence of a static view of development but seems somewhat unlikely. By contrast, in a dynamic approach to development, the role of the infant in its own development and the complex interaction between the infant's behaviour and genes and experience is crucial. In this view, the endstate arises from the developmental process itself and is not already present at the outset. These ideas will be developed further in Chapter 8.

The dynamic approach to neurodevelopmental disorders stresses the importance of characterising cognitive function in infancy. Neurodevelopment plays a role in shaping cognitive function even after birth (Elman et al., 1996). It is crucial to examine cognitive ability from the very outset of development, to investigate whether differences in performance occur as a product of different developmental processes, affecting learning and the processing of inputs from the environment, or whether the differences in neurodevelopmental disorders are already present at birth. It would in fact be advantageous to follow individuals throughout development, using a fully longitudinal design, but that is beyond the confines of a 3-year thesis. This would allow developmental trajectories to be charted to see whether they are similar in infancy or diverge during postnatal development, and precisely when and how this divergence occurs.

In order to apply a developmental approach, two aspects of cognitive development in WS and DS were examined both in infancy and in the phenotypic outcome, although of course in long-term postdoctoral research it will be essential to chart the

developmental trajectory between the two. The domains of number and language were chosen because language abilities differ substantially in the two syndromes in the mature phenotype, but number abilities are reported to be impaired in both clinical groups. This difference in outcomes, both within and across syndromes, allows a number of issues to be addressed.

1.3.1 Aims and questions to be addressed

One: to characterise the cognitive profile of infants and the phenotypic outcome (a group consisting of older children and adults) in two syndromes, Williams Syndrome and Down's Syndrome, using standardised ability tests, in order to compare adult and infant profiles of abilities. The description of the cognitive profile in infancy means that it is no longer necessary to infer infant cognitive profile directly from our knowledge of the adult state. It may well directly challenge what one might call the "Modularity Continuity Hypothesis".

- Is the pattern of strengths and weaknesses in the mature cognitive profile the same as that present in the infant cognitive profile?

Two: to examine the abilities of infants with WS and DS in selected areas of language and number, namely vocabulary and numerosity discrimination, in order to investigate whether the within- and across-syndrome differences in the abilities present in the mature phenotype are present from the outset or whether they arise as a consequence of developmental processes.

- Is vocabulary a relative strength for infants with WS, as it is in adulthood, or does the infant cognitive phenotype differ from that in the endstate?
- Is vocabulary a relative weakness for infants with DS?
- Is number impaired in infancy in both syndromes, as it is in adulthood?

Three: to verify the mature cognitive phenotype for selected aspects of language (vocabulary and syntax) for both WS and DS and also to characterise selected aspects

of number ability in these syndromes. Little is known about number ability in WS, so preliminary studies must be carried out before the developmental trajectory can be examined.

- Are these aspects of language a relative strength in WS and a relative weakness in DS in the mature phenotype?
- How do individuals with DS and WS perform on a selected range of number tasks?

Four: to devise and use tests that enable a comparison of abilities to be made across two very different periods of development. The studies in this thesis employ tests which are as homologous as possible for infants and adults. In the domain of language, receptive vocabulary tests and tests of receptive syntax are used for both the adult and infant groups. For number, infants and adults were tested using a numerosity discrimination paradigm that was presented in an age-appropriate manner.

1.4 Structure of the thesis

A study addressing the first aim of this thesis is presented in Chapter 2. Infants with WS and DS and their controls were tested using the Bayley Scales of Infant Development (II), in order to construct the pattern of strengths and weaknesses in the infant cognitive profile. This was done by contrasting language and non-verbal skills. In chapter 3, a similar procedure was used to verify the cognitive profile in the mature phenotype of WS and DS and control participants, using the British Ability Scales.

Chapters 4 and 5 consist of studies of infant language and number ability respectively. The second aim of this thesis is addressed in these chapters along with consideration of aim 4, the use of homologous infant and adult tests.

In order to compare adult and infant data, it is necessary to verify the adult profile for WS and DS (aim 3). The adult profile is addressed in Chapters 6 and 7 which are reports of studies concerning language and number ability in late childhood and adulthood. These chapters are not the main foci of the thesis but provide data which support the developmental approach taken in the thesis and provide background data for the investigation of number understanding.

Finally, chapter 8 provides a general discussion of all the data. This addresses questions concerning changes in ability across development and whether the impairments and relative strengths observed in the mature phenotypes of WS are DS are present in infancy or are products of different developmental processes.

1.5 The Research Program

Before embarking on the empirical work in this thesis, I will give a brief overview of how this research project was structured and carried out.

1) Visits

Individuals in this research project were seen for one or two sessions. For adults and older children this was either at home or at the laboratory. For infants, at least one session was conducted at the laboratory, with a home visit used only for standardized testing.

During the session infants and older participants underwent all the tests described in this thesis, along with others reported elsewhere (Brown, 2000). Data were gathered for all experiments during the course of one or possibly two visits.

This was done because of the nature of the syndrome under investigation. Williams Syndrome is a very rare disorder, so in order to gather a reasonable sample size it was necessary to recruit participants from all over the United Kingdom. This meant that multiple visits, and the more cumulative type of research possible with such a program was not feasible. Families could not commit to multiple long distance trips and it was not possible to take many of the experimental tasks to them. Instead, testing was done on few occasions but was very intensive, with all assessments carried out on the same visit.

2) Participants

Participants underwent a large battery of tests during their visits. Therefore, each individual contributed data to many of the studies reported in this thesis. Not all

participants contributed to all the studies, but most contributed to a large number of them.

In the tables of individual data, participants are identified by a unique code, so their participation across studies can be traced, if the reader is interested.

2

The overall cognitive profile in infants with Williams Syndrome and Down's Syndrome: Performance on the Bayley Scales of Infant Development II

2.1 Introduction

This chapter focuses on results from the Mental Scale of the Bayley Scales of Infant Development II (Bayley, 1993). The Bayley Scales are widely used by health professionals to screen infants who may be at risk of development delay and to assess the effects of intervention that may be given. Despite the use of the scale with infants with developmental delay, there is very little published work concerning its use with infants with Down's Syndrome and even less for infants with Williams Syndrome.

In this chapter, I will outline the structure of the Bayley Scales and comment on their use with atypically developing infants. Previous research which has employed the Bayley with infants with Williams and Down's Syndrome will be discussed, before describing a study in which the Bayley Mental Scale was used to characterise the cognitive profile of the participant infants. The aim of the present study was to investigate the difference in performance for cognitive and language items in 24, 30 and 36 month old infants.

2.2 The Bayley Scales: A general overview of The Bayley Scales of Infant Development (BSID II)

In order to identify phenotypic differences in the cognitive profile of infants with WS and DS, the Bayley Scales of Infant Development (Second Edition) were used. These scales consist of three standardised sub-scales: the Mental Scale, the Motor Scale and the Behaviour Rating Scale (BRS). The three scales give a comprehensive overview of current developmental functioning of 1-42 month old infants. The Mental Scale is of particular interest for the purposes of the present study and will be discussed in greater depth below. The Motor Scale assesses the control of fine and gross muscle groups, while the BRS is used to assess behavioural indices during the test, such as emotion regulation, engagement and orientation to the tasks and people around, and motor quality. The scores on each subtest can be compared with norms from 1700 typically developing children drawn from a stratified sample of the US Population. One hundred children have been tested in each of 17 age groups, with a greater number of age groups sampled in the 1- 12 month range because of the more rapid development found in younger infants.

The scales are designed to "assess the infant's current level of performance from observation of the infant's interaction with stimuli designed to engage him or her" (Bayley, 1993, p. 2). It is also noted that the scales are often used to identify areas of development that are delayed. Given this, one aim of the revision of the original scales (BSID I) was to improve their clinical utility and validity. BSID II was used with all infant participants here.

2.2.1 The Mental Scale

This chapter is concerned only with the Mental Scale. The items on this scale cover four facets of development: Cognitive, Language, Motor and Social. These can be treated as one, in an overall score, or age equivalents can be calculated for each of the four facets. In order to allow administration of age-appropriate items, the scale is divided into shorter item sets.

The mental scale consists of 22 item sets, each appropriate for testing children in a particular age group. Within an item set, there is a wide range of age-appropriate tasks designed to assess different aspects of cognitive development. These include infant recognition memory, visual preference visual acuity, problem solving, concept of number and counting, language development, and personal and social development. The item sets contain tasks of varying difficulty. The easiest tasks are passed by 90% of the standardisation sample at the appropriate age and the hardest by only 15 %.

The administration of the sets allows for the items to be presented in a flexible order and for the previous set to be administered if the child does not reach a particular level of attainment. If the child does very well, there is scope to administer harder item sets until a ceiling is reached. This method is somewhat problematic when used with atypically developing populations. Chronological age-appropriate item sets would be too demanding for many if not all of these participants. This would mean that the test session would involve the administration of many item sets in order to find a set on which the infant is able to succeed on the necessary number of items. The problem of establishing a starting point with atypical populations has been the topic of much discussion (c.f. Ross & Lawson, 1997 on this problem when dealing with premature infants) and will be returned to in the procedure section of this chapter.

Interpretation of scores for typically developing children

Scores on the Mental Scale can be interpreted in three ways. The raw score is the number of items correct between the basal and ceiling items. This raw score can then be converted into a standardised score, the Mental Development Index (MDI), using the tables provided in the manual. Index scores between 50-150 (approximately 3 SDs above and below the mean) are provided. The majority of the atypical infants tested in the present study could not be given an MDI. The score for these infants is recorded simply as <50. Robinson and Mervis (1996) have now published tables of MDIs below 50, extrapolated from the normal population. They used regression equations to calculate MDIs from 30-50, using data from the lowest scoring group of infants in the standardisation sample.

An age-equivalent score in months can also be calculated from the raw score, although caution should be exercised when interpreting age-equivalent scores. However, such scores are useful when matching groups of subjects.

For a more detailed analysis of performance, it is possible to examine the child's performance over four facets to identify particular strengths and weaknesses. To do this, all the items have been divided into four sub-groups: cognitive, language, motor and social. The last of these is problematic due to the small number of items and the fact that no items are given above the 9-month level. Items completed are marked off for each facet and a visual profile of performance emerges. The child is said to be performing at a level where "mastery" is achieved, i.e., where a predominant number of items are passed. So, for example, a child may perform at the 9-month level on language, at 12 months on the cognitive facet, and at 11 months on the motor facet.

2.2.2 Previous research using the BSID with Williams Syndrome and Down's Syndrome

Very little research on the performance of infants with Williams Syndrome or Down's Syndrome on the Bayley Mental Scale has been conducted. The little that has been done with infants with Down's Syndrome tends to focus on the level of stability of their performance between test and retest, to which I now turn.

Down's Syndrome

In order to investigate the stability of performance for infants with Down's Syndrome, Wishart and Duffy (1990) administered the Bayley twice to 18 infants and children with DS ranging from 6-48 months, with an interval of between one and two weeks between each session. They found that despite consistent raw scores from test to test, the underlying pattern of performance on individual items changed considerably, suggesting that item-to-item stability measures are very important. The pattern of performance differed from test to test in two ways. First, using predictions on the

basis of standard errors of measurement they found that variability of performance found in DS subjects was different from the variability found in normal subjects. Second, the patterns of fails-to-passes and passes-to-fails differed. The authors identified a significant difference between the predicted variability and the actual variability in scores between test sessions. There were in fact 165 cases of instability when the items for each infant were examined, with 55% of these changing from a pass to a fail and 45% changing from a fail to a pass. If an infant passed an item that it subsequently failed, then it is very likely the item was within the infant's ability and that the pass was genuine. The second failure can probably be explained by motivational or attentional problems, both of which are known to occur in Down's Syndrome (Wishart, 1987). Changes from fail to pass are also likely to be exacerbated by these factors. It is rather unlikely that the infants will have suddenly developed the ability to pass a great many more items in under two weeks, thus ruling out an argument on the grounds of sudden leaps in performance due to interim development. The infants in the Wishart and Duffy study were also familiar with the experimenter before the test session, so it is implausible that improved performance was due simply to increased familiarity with the test administrator. In their analysis, the authors classified the infants' failure to engage in three ways: refusal to attend, rejection of the objects, and repeated production of inappropriate off-task behaviour. These failures to engage accounted for 62% of pass-to-fail items and 48% of fail-to-pass items, again adding to the evidence that attentional factors may contribute to variable performance on developmental tests in young children with DS.

Another study investigating the stability of performance in infants with Down's Syndrome produced different results. Wright (1998) found that the pattern of passes and fails on the Bayley Mental Scales of infants with Down's Syndrome followed a similar developmental sequence to that seen in typically developing infants. In addition, contrary to the findings of Wishart and Duffy, Wright discovered that the stability of DS infants' performance was relatively high and comparable to the normal control group over a short period (four weeks). Both groups' mean level of consistency was around 83%. Moreover, even over a longer test-retest interval (8 months), consistency still remained high. Stability of performance in DS has also

been reported by Messer and Hasan (1992). In sum then, it is possible that the performance of infants with DS on the Bayley may be quite unstable, although there is some disagreement about this in the literature. The possibility of instability in the performance of both infants with DS and WS should be considered when evaluating Bayley scores for these infants.

In the following section, evidence for a difference between the cognitive profiles of infants with DS and WS will be considered.

Comparisons between Williams Syndrome and Down's Syndrome

Mervis and her colleagues (1997) compared the cognitive performance of 6 infants and toddlers with Williams Syndrome and 6 with Down's Syndrome, based on the first edition of the Bayley Mental Scale (BSID). The children were seen at approximately 6 monthly intervals, ranging from 6 to 30 months. Items on the scale were divided into cognitive or language items. Scoring began with the tenth item below the basal (first item failed) and continued to the point where testing was discontinued. The proportion of cognitive items passed from all those attempted, as well as the proportion of language items passed from all those attempted, was calculated, and mean proportions were reported for each group.

Mervis reported that the children with Williams Syndrome exhibited the same pattern of language and cognitive performance as seen in the adult WS phenotype. They performed better on language than on cognitive items, even at this early stage. The converse held for the group with Down's Syndrome. They did better on cognitive items than on language items. This is what one would expect if performance in infancy and early childhood could be directly inferred from the mature phenotype. However, these data emanate from a rather small group of subjects, and the methods used to calculate the language and cognitive scores are not explained clearly. These issues will be considered in greater depth in the discussion section.

2.3 The present study

Given that the adult literature suggests that both Down's Syndrome and Williams Syndrome have distinctive cognitive profiles, the present study will use the BSID II, with its items tapping cognition and language, to examine whether these profiles are present in infants with DS or WS.

The adult profiles will be considered briefly here, to provide a benchmark for infant performance. Individuals with Williams Syndrome are reported to perform poorly on tests which assess non-linguistic cognitive ability, such as those measuring visuo-spatial skills like block design (Bellugi et al. 1988,1992,1994; Bertrand et al., 1997), but do relatively well or equivalently to their MA on items tapping language skills (Bellugi et al., 1988,1992,1994). For Down's Syndrome the profile is less uneven, but individuals display poor performance on language tests and rather better performance on measures of non-linguistic ability (Fowler, 1990; Rosenberg & Abbeduto, 1993; Wang & Bellugi, 1993). This is the opposite of the pattern seen in WS. Mervis et al. (1999) have identified these contrasting profiles using a single test, the Differential Ability Scales, the American version of the British Ability Scales (Elliott, 1990).

Given this background, which suggests that the cognitive profiles of individuals with WS and DS differ in the mature phenotype, the present study aims to investigate whether the patterns of strength and weaknesses evident in each syndrome are present from the outset of development in infancy. If the patterns present at this early stage are the same as those in the steady state, this would suggest that the difference in cognitive phenotype is present from the very beginning of development. By contrast, if the patterns found in infants differ from those in adults with the same syndrome, then these differences would not be present from the outset, but would stem instead from different developmental trajectories in each syndrome.

This study will characterise the cognitive profile of each group of infants, using the Bayley Mental Scale II. Results will be discussed in the light of Mervis' research and in relation to the pathway from infancy to the mature phenotype.

2.3.1 Method

Participants

71 infants took part in this study, yielding 81 cases. They were seen as part of a much broader study investigating the cognitive development of infants with Down's Syndrome and Williams Syndrome. Infants were seen as close to 24, 30 and 36 months as possible. Some infants were seen only once, others twice. Fifteen infants with Williams Syndrome were tested. Six were tested twice (at an interval of 6 months) and nine were seen once, yielding 21 tests. Twenty infants with Down Syndrome were tested. Four were tested twice (again, at an interval of 6 months) and sixteen were seen once, yielding 24 tests. In addition, 19 MA-matched normal infants and 17 CA-matched normal infants were tested. The mean ages and standard deviations for each group are given in Table 2.1 below:

Table 2.1 Mean Chronological and Mental Ages for each group.

	Mean age (months)	SD	Range (months)	Mean mental age (months)	SD	Range (months)
WS	30	4.56	24-36	16.5	2.39	14-21
DS	29.5	4.87	24-36	15.3	2.32	12-20
MA-matched	15.6	2.42	12-20	15.3	2.55	11-21
CA-matched	30	5.24	24-36	30.6	5.14	25-40

Procedure

Establishing a starting point

In order to test the atypically developing subjects on an appropriate item set, it was necessary to establish a standard procedure for choosing the item set with which to begin testing. Had testing begun with the set appropriate for the child's chronological age, then it would have been necessary to move down several times to a lower item set before a basal could be established. To avoid this unnecessary increase in the length of the testing session, it was decided that all the atypical children would be assigned a starting item set according to a standard proportion of their chronological

age which might reflect their level of cognitive functioning. This was done by assessing findings in the existing literature. Reports suggest the young children with Down's Syndrome perform at a level approximately one half to two thirds of their chronological age on a variety of measures (Carr, 1985; Wishart & Duffy, 1990). Data on vocabulary development, grammar development and IQ measures all support this conclusion. It was therefore decided to start testing each child on the item set closest to half his or her chronological age, choosing sets that formed an uninterrupted sequence. This meant that for all children with DS and WS, testing for 24 month olds began with the 13 month item set, testing for 30 month olds with the 14-16 month set, and testing for 36 month olds with the 17-19 month set. Apart from the choice of starting set, testing proceeded according to the manual. The child could, if necessary, progress onto a higher item set or be moved down to a less demanding one.

The test session

The majority of infants were tested in their own homes. This session was normally conducted after the infant had visited the laboratory to take part in experimental studies. A minority of infants were tested during their visit to the laboratory, as part of a longer assessment of language and cognitive development. The administration of the Bayley usually took place the afternoon before the principal testing session. Test sessions lasted approximately one hour, with breaks incorporated where necessary. When possible, infants were tested at a small table, seated opposite the examiner. On a few occasions however, testing had to be carried out on the floor.

The Mental Scale was administered in accordance with the instructions given in the manual, with the exception that, as described above, testing for atypically developing infants began with a lower item set than that appropriate for their chronological age. Both the chronological age-matched and mental age-matched controls were tested beginning with the chronological age-appropriate item set, as instructed.

Scoring

As mentioned in the introduction, it is possible to score the Mental Scale in three ways. First, the infant's raw score can be converted into a standardised developmental index or MDI. Second, a mental age equivalent can be given or third, an age equivalent on each of the four facets can be ascertained. The majority of infants with Down's or Williams Syndrome did not achieve a high enough raw score to be given an MDI and were therefore designated MDI <50. A mental age equivalent was calculated for each infant and this was used for subsequent mental age matching across syndromes and to the typically developing controls. The mental ages taken from each separate facet were not used for either matching or analysis in this study for a number of reasons. Firstly, the social facet has very few items, which means its interpretation is problematic. Secondly, the scoring criterion for assigning a mental age for each facet is unclear. A mental age score would also not be as accurate as a quantitative measure when comparing performance on particular aspects of the scale. One child may be classified at the 13-month level using the facet method having passed 6 out of 10 possible items, whereas another infant may score 9 out of ten and yet be given the same classification. Therefore, for the purposes of this study, to compare language and cognitive functioning in the four groups of infants, analysis was carried out on the basis of raw scores.

2.3.2 Results

The relative difference between cognitive and language scores

Two scores were calculated for each infant: the proportion of language items correct and the proportion of cognitive items correct. This allowed for the analysis of the relative difference between language and cognitive performance for all individuals irrespective of their overall performance on the BSID II. Language items were taken from the language facet of the Mental Scale. Cognitive items were those on the cognitive facet. If an item appeared on both the cognitive and the language facet, it was assigned to the language facet only. This was done because all items on the

language facet also appeared on the cognitive facet. The proportion of items correct was calculated as follows and converted into a percentage:

$$\text{Proportion} = \frac{\text{Number of items correct (all items prior to the item set + items passed in the set)}}{\text{Number of items administered (all items up to the end of the tested item set)}}$$

This enabled the infant's performance on cognitive items to be compared with his or her performance on language items. Table 2.2 shows the mean percentage of language and cognitive items correct for each group. Readers interested in individual data can find this tabulated in Appendix B.

Table 2.2 Mean percentage of cognitive and language items correct for each group

	N	Mean percentage of cognitive items correct	SD	Mean percentage of language items correct	SD
WS	21	92.38	4.57	84.61	11.55
DS	24	91.97	4.08	75.85	10.43
MA-matched	19	92.85	3.68	69.16	9.78
CA-matched	17	94.95	2.60	87.39	5.07

The percentage of language items and cognitive items passed by each infant was entered into a repeated measures Anova, with group as the between-subjects factor (WS, DS, MA-matched, CA matched) and type of item (language or cognitive) as the within-subjects factor¹. An effect of item type emerged, with all the infants performing better on cognitive items than on language ones, $F(1,77) = 140.47$, $p < .0001$. Post hoc t tests were carried out to examine these differences in each group. The results are reported in Table 2.3 below. A significant effect of group was also found, $F(3,77) = 13.89$, $p < .0001$. As would be expected, CA-matched infants did better than the other three groups on both language and cognitive items. In addition, there was an interaction of group by item type $F(3,77) = 10.54$, $p < .0001$, suggesting

¹ Some participants contributed more than one data point to the analysis. Since the statistical analysis assumes independence of data points, any correlation between an individual's two data points may reduce the significance of the result. However, to minimise this problem, these data points were collected with an interval of at least six months. This has been done in previous research with infants with rare neurodevelopmental disorders, e.g (Mervis et al. 1999).

that performance across cognitive and language items varied according to the group to which the infant belonged.

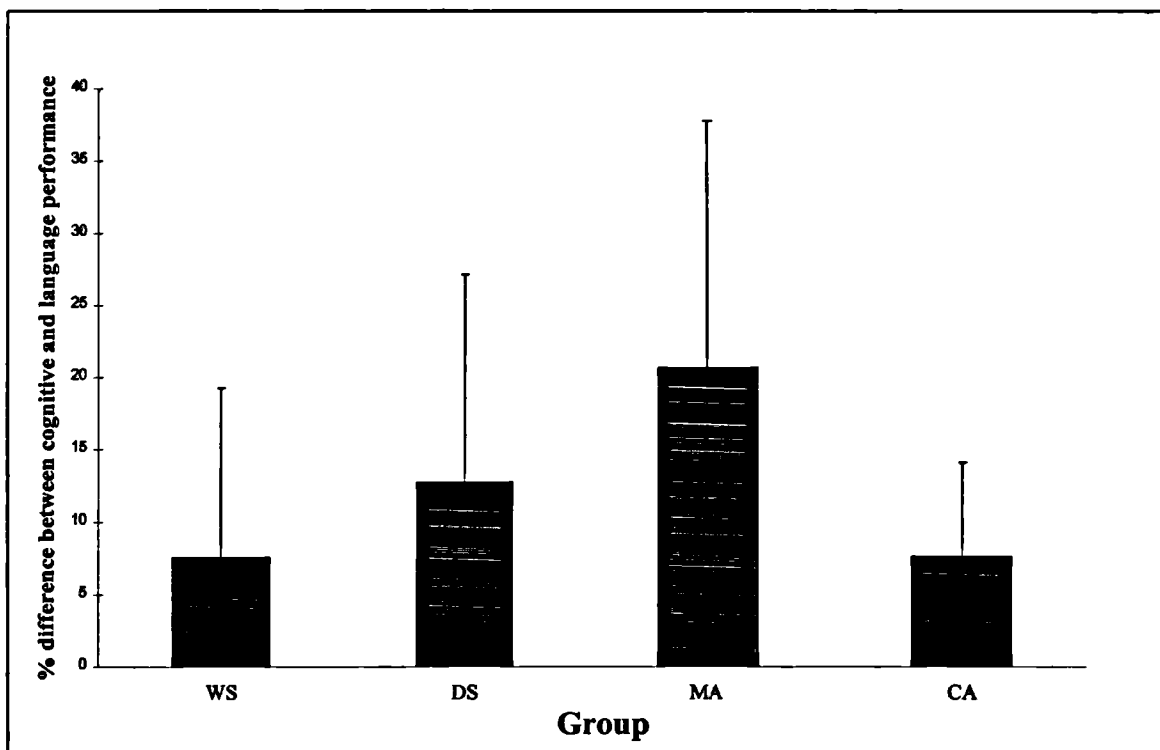
Table 2.3 t tests examining the difference between percentage of cognitive and percentage of language items correct for each group

	t value	df	p value
WS	2.96	20	.008
DS	6.79	23	.000
MA-matched	10.24	18	.000
CA-matched	5.78	16	.000

In order to interpret this interaction further, one way Anovas with group as the between-subject factor, and either language or cognitive performance as the dependent variable, were carried out. A simple main effect of group was found for performance on the language items, $F(3,77)=13.83$ $p<.0001$, but not for cognitive items, $F(3,77) = 2.19$, NS. Therefore, the differences between performance across the groups reside in their language performance. Post-hoc analysis of language performance revealed significant differences between the Down's Syndrome (mean 75.85) and Williams Syndrome group (mean 84.61) Tukey HSD, $p<.05$. The Williams Syndrome group differed from all groups except the CA-matched controls. It should be stressed at this point that performance is measured as a proportion of items that were attempted, so that results between groups are relative patterns, not absolute similarities or differences. The pattern seen in the DS group is like that of the MA group, with no significant differences in cognitive or language performance, whereas the WS group pattern resembles that of the CA group. This suggests that the DS group's performance is delayed. The infants with DS are performing like younger typically developing infants, with a large discrepancy between their cognitive and language abilities. On the other hand, the WS group exhibits a pattern similar to that of children of their actual age but is actually performing at a much lower level than the controls and on a completely different set of items. In contrast to the DS group, the WS infants can therefore be said to exhibit a deviant rather than delayed pattern of abilities. Figure 2.1 illustrates this difference between language and cognitive items for each group.

In contrast to Mervis et al.'s (1997) results, in the present study the difference between cognition and language for the DS group is larger than that present in the WS group and goes in the same direction as observed in WS, rather than the opposite. In infancy then, all groups -typical and atypical- perform better on cognitive than language items. Furthermore, it is noteworthy that DS infants show a larger discrepancy, or more uneven profile than WS infants. This is the opposite of the pattern present in the mature phenotype.

Figure 2.1 The mean percentage difference between cognition and language performance for each group



Changes in the profile with age: cross-sectional data

Given that I had collected cross-sectional data from infants at 3 different ages, an analysis of variance was carried out to determine whether the difference scores for each group change with age. Difference was the within-subjects factor and age and group was the between-subject factors. The MA-matched group was excluded from this analysis because the infants did not fall into the 24, 30 and 36 month categories

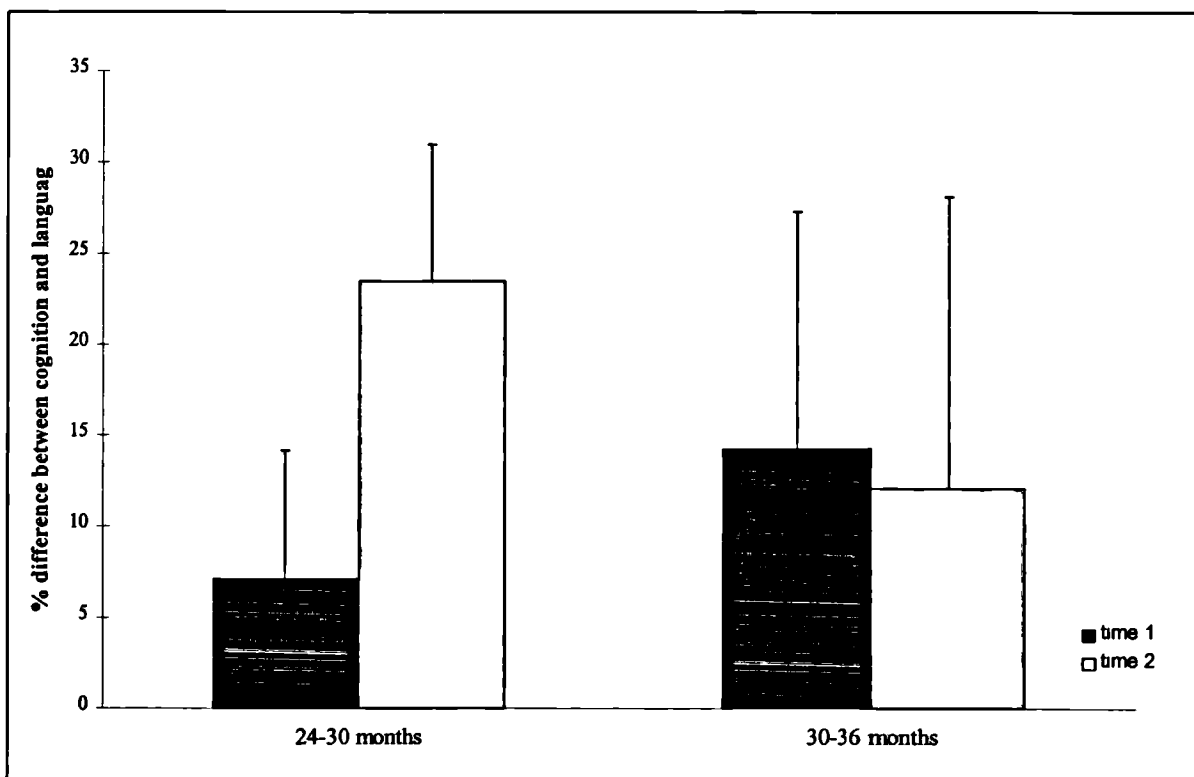
which made up the other groups. An effect of age on difference scores, $F(2,61) = 4.63, p < .01$ and interaction of age by group was found, $F(4,61) = 6.19, p < .0001$. Further analysis of the interaction, looking at simple main effects, revealed that difference scores were affected by age in both the DS, $F(2,23) = 18.15, p < .0001$ and CA-matched group, $F(2,16) = 10.04, p < .01$, but not the WS group, $F(2,20) = 1.79, NS$. Post hoc analysis showed that these differences were due to a developmental change between 24 and 30 months in both groups. In the DS group, the difference increased from 24 to 30 months, from a mean of 3.30 to 22.59, Tukey HSD, $p < .05$. In the CA-matched group, the difference decreased from 12.83 to 6.08, Tukey HSD, $p < .05$. In other words, the difference between cognition and language decreased in the CA group between 24 and 30 months, whereas in the DS group the reverse was true. This is probably because the language delay seen in DS infants becomes more pronounced as development progresses. The Bayley Scales demand more language ability with each increasing item set and therefore the gap between language and cognitive performance can widen. The CA-group shows a decreasing difference because their language skills are catching up with their cognitive skills, as one would expect in normal development. The difference in the WS group from 24 to 30 months was not significant. It should be noted that these data are from a cross-sectional design and so cannot necessarily shed light on individual patterns of development. In the following section, a brief consideration is made of some longitudinal data which can begin to characterise the developmental profiles of individual infants.

Changes in the profile with age: a longitudinal aside

In this study, 4 infants with DS and 6 infants with WS were seen on two occasions, six months apart. It is therefore possible to offer some conjectures on the nature of the phenotypic cognitive profile over time. However, it is important to note that the sample size for this part of the study was small and so the data should be taken as preliminary. Figure 2.2 shows the difference at two time points between the proportion of language and cognitive items correct for DS and WS infants. The pattern of strengths and weaknesses, as measured by the difference between cognition and language, did not change over this time for either group, $F(1,8) = .98, NS$. However, if the data are divided into two age groups, those from 24-30 months and

those from 30-36 months, then the patterns change. A repeated measures Anova, with age group as the between-subjects factor and difference at time 1 and time 2 as the within-subjects factor, revealed an interaction between time group (24-30 or 30-36 months) and scores at time 1 and time 2, $F(1,8) = 9.20$, $p < .05$. There was a significant difference between difference scores at 24 and 30 months, $t(2) = -18.9$, $p < .01$, but not at 30 and 36 months, $t(6) = .57$, n.s. This is an interesting result, given the findings from the exploration of cross-sectional data reported above. The longitudinal data fit with the source of the difference in the cross-sectional results, suggesting that there are important changes occurring in the pattern of cognitive and language abilities between 24 and 30 months. Of course, the longitudinal data come from a very small group and so may not be entirely representative of the populations as a whole. Therefore, a replication with a larger group of infants is needed before conclusions can be drawn. Control infants were not seen on more than one occasion so this analysis could not be carried out for the typically developing group.

Figure 2.2 The difference between the proportion of language and cognitive items correct for DS and WS infants over time



2.3.3 Discussion

Cognitive performance is better than language performance for all groups.

Williams Syndrome

This result is different from Mervis' 1997 findings, in which her WS group performed better on language than cognitive items, even at this early stage of development. Mervis' results are somewhat surprising, given the age of her participants. Despite relatively good language ability in later life, language delay is a major marker of Williams Syndrome. Infants with WS begin to produce language considerably later than their typically developing counterparts (Thal, Bates & Bellugi, 1989). Therefore, it seems unlikely that at a stage at which a child with WS would be producing little or no language, their language skills would be more markedly developed than their cognitive abilities. In the present study, all infant groups found the language items challenging. Many parents commented that their children were not yet labelling or identifying objects when these tasks were presented to the child as part of an item set. These parental assessments of their infants' ability to complete an item should be taken seriously, as several studies have shown that parents' predictions of their infant's performance on the Bayley are remarkably accurate (Karraker & Coleman, 1999).

Other evidence that supports poor WS performance on language items is also available. Several of the core language items on the Bayley Mental Scale employ pointing as a means of response for the infant. This could contribute to the poor performance of infants with WS because pointing is a particular problem area for these infants. Infants with WS do not follow the normal developmental pathway for pointing behaviour. Typically developing infants point out objects in their environment as a means of communication well before they begin to produce language. In WS, this is not the case. Pointing behaviour emerges well after production begins (Mervis et al., 1997). If, indeed, all WS infants do follow this pattern, then many of the infants would not be expected to be using referential pointing at the time of testing and would therefore be at a disadvantage on many of the language items. For example, those with instructions like, "Point to the Dog,

where's the dog?" or others such as "Where's your nose? Show me your nose?" When parental assessment of the test session was sought at the end of the Bayley, many parents of WS infants noted that their child never pointed to pictures spontaneously or on command, as we had expected of them. These findings suggest that pointing difficulties may be a valid explanation of poor performance on some of the language items. It is surprising, however, that the infants in the Mervis study did not have the same difficulties with language. This difference is discussed in a later section (starting on page 49).

Down's Syndrome

The DS group was expected to be poorer at language than cognitive items, and indeed this was the pattern that emerged. The pattern found in the mature phenotype, i.e., better general cognition than language performance, also holds in infancy. As seen above, this is not the case for Williams Syndrome.

MA-controls and CA-controls

In normal development, language ability lags behind cognitive ability in early childhood. It is, therefore, not surprising that in the control groups language performance was poorer than cognitive performance. However, the MA group did extremely poorly on language items. The MA matched infants in this study, who had a mean age of 15 months, were in the very early stages of language production. This is supported by data from a study of vocabulary acquisition using a parental report measure called the MacArthur Communicative Development Inventory (M-CDI) (Hamilton, Plunkett & Schafer, 2000). This study found that at the age of 16 months infants in the UK are able to understand around 112 words and to produce around 6 words. This is well below the level expected for an infant in the USA, where standard performance on the M-CDI was 200 words produced and 37 understood at 16 months. With this in mind, note that the Bayley is standardised using an American sample. It is thus likely that expectation of language level would be higher for the test than that found in the general UK population. To illustrate this, the Bayley item set for 15-month age group contains many language items. Infants are expected to identify body

parts, produce two words spontaneously and both name and point to pictures. The infants in the present study did particularly poorly on the items which tapped spontaneous language development, e.g., combining word and gesture and producing two words appropriately. Some of the infants may have been able to do these, but were perhaps unwilling to do so in the presence of a relatively unknown adult. However, as mentioned above, many parents noted that their infant was not yet at this stage of development. Noteworthy is the fact that item sets in the Bayley Scales are designed so that a smaller proportion of infants pass items at the end of the set than those who pass items at the beginning. If the majority of cognitive items fell towards the beginning of a set and the language items towards the end of a set, then language items would be at a disadvantage. Indeed, six of the language items in the 14-16 month item set were the last items of that set. In order to investigate this idea further, the first five and the final five items for each item set used in this study were examined. It was found that there were more language items in the final items (mean 3.12) than in the first items (mean 1.67) and that performance for all groups was worse for the final items (mean number incorrect 3.25) than the first items (mean number incorrect 1.24), as suggested in the Bayley manual. This is a shortcoming of the Bayley.

Differences in language performance

The differences in the cognitive profiles of the four groups were caused by performance on language items. The performance of the DS group was particularly poor. Language difficulties, particularly problems with language production, have been widely reported in the DS literature (Chapman, Seung, Schwartz & Kay-Raining Bird, 1998) and results from the MacArthur Communicative Development Inventory, an early vocabulary measure, suggest that the disadvantage seen for production in older individuals with DS is already present in infancy. Both the WS and DS groups are poorer at producing than understanding words, but the difference in the DS group is far more pronounced. The difficulties that individuals with DS have with language production have been attributed to several factors, including hearing deficits, poor verbal memory and problems with articulatory mechanisms (Chapman et al., 1998). In

Chapter 4, language abilities of infants with WS and DS are discussed in greater depth.

Changes in cognitive performance with age: cross-sectional and longitudinal findings

Using both a large cross-sectional sample and a small sample of the same infants at two time points, it was found that the size of the difference between cognitive and language performance did not change between 30 and 36 months. The major change occurred between 24 and 30 months. This period, between two and two and a half years, is a period of crucial development in infants with DS and WS. Over the testing period, this was the time at which the experimenters noticed the greatest change in the infants. In the control infants, the difference between language and cognitive skills decreases. This is expected as language abilities catch up with non-verbal skills. However, in infants with DS this difference actually increases. It is likely that the problems infants with DS have with language become more pronounced with age, as the demands placed upon them are greater. At present, it is not possible to comment on changes in WS in great depth. In the large sample, no differences were found with age for this group. In the longitudinal sample, a similar difference was found for WS as for DS infants but it must be noted that the longitudinal sample was very small.

In summary, then, it seems that there are some changes over time in the profiles of WS and DS infants who were followed longitudinally. However, future research will require a larger longitudinal sample to explore this further.

Differences between results from Mervis et al. (1997) and the present study

Given the results reported here, why did Mervis and colleagues (1997) find not only a larger difference between language and cognitive items in WS, but also a difference in the opposite direction, with language scores higher than cognitive scores. In my view, the results differ for two main reasons: 1) the use of the BSID I instead of BSID II in the Mervis et al. study and 2) the sample of infants tested.

BSID I versus BSID II

Mervis used the first version of the Bayley Mental Scale to produce her cognitive profiles. This differs in several ways from the second version. First, there are no facets on the first version and therefore Mervis allocated language and non-language items using her own personal criteria. These items are not clearly specified and may differ from those taken from the facets used in the present study. Second, BSID I, has no item sets. This means that the number and type of items included in the analysis in the two studies differ and would result in different proportions of language and non language items passed for each study. Mervis used the following criterion: items beginning with ten items below the first item failed until the end of testing would be used in the calculation of proportions. By contrast, in the present study all items prior to the basal and those items passed within the item set were used in the calculation. The differences in methodology may well have led to the different outcomes of these studies. In order to settle this empirically, future research should aim at testing the same infants twice, using both the old and new versions of the Bayley.

The samples studied

In the present study, scores were included from at most two test sessions for each infant. Five infants with WS and four infants with DS were seen twice, while all other infants were seen once. This is not true of the infants in Mervis' study, which was longitudinal. Mervis tested each infant on average four times. This meant that each infant contributed 4 of the 24 data points for their syndrome, either DS or WS. Overall, then, Mervis had data from only 12 infants, whereas for the present study DS and WS data came from 35 infants. This may have been another contributing factor to the discrepancy between the studies. If only some of Mervis' WS infants exhibited the greater language than cognitive ability pattern, then this would have contributed to the overall mean on four occasions. By contrast, in the present study the profile of a wider range of infants contributed to the mean cognitive profile. Therefore the effects of one or two infants with a particular syndrome exhibiting the mature phenotypic pattern were not able to overshadow the pattern emerging in the group as a whole. Furthermore, data from the present study suggest that the profiles of infants change

over developmental time between 24 and 30 months. While Mervis and colleagues tested infants up to 30 months old, they did not carry out developmental analysis, although the data would have allowed it.

In order to tackle these discrepancies, it is necessary to carry out careful longitudinal testing of a much larger sample of infants, something that was not possible within the scope of the present research.

2.4 Problems with the Bayley Scales as an indicator of cognitive profile

Despite the use of the Bayley Mental Scale (version II) for the present study, it is important to note that there are many problems with the use of this test with atypical populations. The most important of these is the choice of a starting point, an issue which was addressed in the introductory section of this chapter. Previous research has highlighted the influence of the starting point on the final raw score of an infant. Nellis and Gridley (1994) provide a very clear example of this, which is illustrated below.

Consider the following: An infant starts on the 6 month item set but fails to establish a basal, getting only 3 credits in that set. Therefore, the 5-month set must be administered. In this set, too, she is credited with too few items to establish a basal, scoring only 4. This means the 4-month item set must be administered. In this set, a basal is finally established. When the infant's raw score is calculated the correct items in the present set (4 month) and those from the two sets above it are added to the basal score (the number of items below the 4 month set). This means that the infant is credited with 7 more items (3 from the 6 month set and 4 from the 5 month set) than she would have been, had she begun simply with the 4 month set from the outset.

Given that choosing a starting point with abnormal infants cannot be done on the basis of chronological age, occasions may arise when some infants are started immediately in a lower item set and so miss out on extra credits gained from starting too high and

working down. The opposite can also occur. Such discrepancies can make a considerable difference when calculating the MA equivalent of a raw score. This becomes even more serious when making comparisons across syndromes. For example, in my present work with Down's Syndrome and Williams Syndrome, it was found that one infant starting in an item set one month higher than another infant, but of the same chronological age and with the same subsequent raw score, had an advantage of 3 months on the language facet, despite no overall difference in language ability when an in-depth item-by-item analysis was performed. Wishart and Duffy (1990) and Oates (personal communication) also argue that different starting points can lead to seriously inconsistent final scores and considerable difficulty with MA-matching. However, in the present study, the use of proportional scores should have surmounted this problem to some extent.

In addition to the "starting point" issue, there are also more general problems with the test. These will be illustrated with evidence from the DS literature because there is no published research concerning these issues in WS. However, it is very likely that similar arguments will hold for infants with WS. It is probable that motivational and attentional factors play a far greater role in the performance of infants with WS and DS than in typically developing infants on the Bayley, yet again, highlighting the problems inherent in MA-matching procedures. The administration of the BSID II is lengthy, which creates an additional problem when dealing atypical infants whose attention span is short. The test manual states that testing takes approximately 25-30 minutes with infants under 15 months and up to 60 minutes for those over 15 months. However, these times are given for normally developing infants. Testing with abnormal subjects can take much longer. For example, infants with Down's Syndrome are very prone to throwing the test items on the floor, if they are not interested in the task, which interrupts and prolongs the session. This disruptive throwing behaviour is seen even in their free play and again occurs far more often than in normal infants (Krakow & Kopp, 1983).

It is also to be noted that infants with DS have problems with attention in all situations and so may not show their true competence on Bayley items. Many of the items on

the BSID II require the infant to interact with the materials spontaneously. This is true, for example, with the cube items. In these items, cubes are placed on the table one by one and the infant is expected to reach for and pick up one cube with each hand, and to try and get a third by some other means (Item 75). This is a particular disadvantage for infants with DS because they spend a great deal of time looking at objects before they engage in a task (MacTurk, Vietze, McCarthy, McQuiston, and Yarrow, 1985). For items such as the cube sequence, the DS infant may be engrossed in looking at the items, and may not think of manipulating them. Reliance solely on visual attention may well be the cause of many failures to engage in motor manipulation.

Inappropriate off-task behaviour may be used as an avoidance strategy. Wright (1998) notes that many infants with DS will avoid items on the Bayley by engaging in social interaction, such as trying to make the examiner laugh or repeating behaviours of which they are capable. It is difficult to decide whether this is related to motivation alone, or to task difficulty. Indeed, Wishart and Duffy (1990) found that some of the problems with engagement might well have been due to task difficulty, which in turn leads to motivational loss. More pass-to-fail items occurred above the midpoint of difficulty for the items attempted by a particular infant, whereas fail-to-pass items were evenly spread. Infants may refuse to participate because they anticipate that tasks will indeed be too difficult. In contrast to normal infants who find difficult items stimulating, infants and children with DS are particularly reluctant to attempt tasks just outside their competency. In addition, they are also poor at consolidating their previous learning (Wishart, 1987,1988). Thus, tasks that they may have recently mastered soon become forgotten. This latter problem may also contribute to the pass-to-fail pattern.

2.5 Chapter Summary

The results of this study highlight the importance and difficulties of characterising the cognitive profile of atypical groups in infancy. Despite the evidence that the cognitive profiles of individuals with Down's Syndrome and Williams Syndrome differ in

adulthood (Mervis, Morris, Bertrand & Robinson, 1999), this study has shown that these patterns are not present in infancy. Both groups perform better on cognitive items than on language items in infancy, with a greater discrepancy seen in Down's Syndrome than in Williams Syndrome. This does not appear to be the case in the mature phenotype where the Williams Syndrome profile is often more uneven than the Down's Syndrome one. Language performance is more frequently higher than cognitive performance in WS adults. This is illustrated by an analysis of data from my own sample of older children and adults with DS and WS which is presented in the following chapter.

The differences between DS and WS in infancy highlight the fact that it is very important not to infer the infant profile directly from the adult pattern. Examining abilities in infancy enables one to investigate whether differences between syndromes occur as a product of development, because of different experience and learning mechanisms, or are present from the outset. If one takes a snapshot of functioning during the steady state, insights into the differences in the developmental trajectories of each group will be lost.

3

The overall cognitive profile in older children and adults with Williams Syndrome and Down's Syndrome: Performance on the British Ability Scales.

3.1 Introduction

In Chapter 2, I demonstrated that infants with Williams Syndrome and Down's Syndrome do not conform to the cognitive profiles that one would expect on the basis of the adult profile reported in the literature. The study reported below aimed at verifying in more depth the cognitive profiles of older children and adults to ascertain whether adult performance in these two syndromes really does conform to classic steady state phenotypic profiles.

Many studies report the presence of a distinctive cognitive profile in adults with Williams Syndrome (e.g. Wang & Bellugi, 1993; Howlin, Davies & Udwin, 1998; Mervis, Morris, Bertrand & Robinson, 1999). The studies suggest that the profile is uneven. Individuals with WS display strong performance on verbal items, such as receptive vocabulary or verbal similarities (Arnold, Yule & Martin, 1985; Udwin, Yule & Martin, 1987). Their performance is much poorer on items tapping visuospatial constructive ability, such as block design (Arnold et al., 1985; Udwin et al., 1987; Howlin, Davies & Udwin, 1998). This pattern does not characterise the DS profile, which is often said to be flatter with all abilities at about the same level (Hodapp, Leckman, Dykens, Sparrow, Zelinski, Ort, 1992; Mervis et al., 1999.) When an uneven pattern is found in the DS profile, then the strengths and weaknesses

are the opposite of those seen for WS. In DS, visuospatial ability can be higher than verbal ability (Wang & Bellugi, 1993; Rosin, Swift, Bless & Vetter, 1998).

In order to characterise the cognitive profile of both groups, the British Ability Scales were used (BAS-II, Elliott, Smith, McCulloch, 1996). These were chosen because their American equivalent, the Differential Ability Scales, were employed by Mervis et al. (1999) in a previous study on the basis of which several claims have been made. The structure of the scales allows for the assessment of abilities in relative isolation, with standardised subtests, so that a pattern of strengths and weaknesses can be constructed. In addition, they provide an overall level of ability. Their structure also allows for the comparison of adult performance with infant performance on the BSID II which is structured similarly. The use of the BAS scales and other standardised tests in previous studies with Down's Syndrome and Williams Syndrome groups is discussed below.

3.2 Previous studies comparing Williams Syndrome and Down's Syndrome cognitive profiles

There is only one published study that uses a standardised cognitive test to compare the cognitive profiles of children and adults with Down's Syndrome and Williams Syndrome. This was conducted by Carolyn Mervis and colleagues using the DAS (Elliott, 1990). 50 individuals ranging in age from 3;11 to 46 years were assessed. The authors conclude that the expected pattern of strengths and weaknesses for WS is present in their sample. Mervis et al. constructed various criteria, using subscales, which reflected the hypothesised profile of WS. They termed this the WSCP or the Williams Syndrome Cognitive Profile. The performance of their participants was then judged against these criteria. 47 of the 50 WS participants conformed to the WSCP, whereas a group of individuals with learning difficulties of mixed aetiology, matched on CA, did not. Despite the size of Mervis' sample, her study has a few shortcomings and different aims from those of this thesis, which prompted a replication in the current study. First, Mervis' study encompassed a wide-age range, which included children who were still developing and adults who had reached the

steady state. However, the current study examined only older children and adults, to look at the profile in the steady state in order to make a comparison with infants. This was not possible in the Mervis study. In addition, Mervis compared her WS group to a mixed aetiology group. In the current study, a comparison of WS and DS was planned to examine these specific phenotypes.

3.3 The present study

The present study differs from Mervis et al. in several respects. Mervis compared her WS group with a mixed aetiology sample, some of whom had Down's Syndrome. This approach does not allow a direct comparison of the Down's Syndrome and Williams Syndrome cognitive profiles, but will only permit separation of WS from other causes of learning difficulties. My study will enable a direct comparison between DS and WS groups to be made, allowing for investigation of differences between phenotypic profiles. A second difference is the creation of new criteria against which fit to a possible DS cognitive profile can be judged. This allows for characterisation of DS in its own right, and not merely in comparison with WS.

It is predicted that if previous findings using several tests are correct, then there will be a distinctive WS cognitive profile which can be derived from a standardised ability test. Individuals with WS should conform to the WSCP, whereas DS participants should not conform. By contrast, DS subjects should conform to a new set of DS criteria (DSCP), if data from previous research are replicated.

3.3.1 Method

Participants

Eight individuals with Williams Syndrome were tested. Seven were tested twice (at an interval of greater than 6 months) and one was seen once, yielding 15 cases. They were matched on sex and chronological age to 9 participants with Down's Syndrome, yielding 9 cases. The mean ages for each group are shown in Table 3.1 below.

Table 3.1 Mean ages of WS and DS groups

	Mean age	SD	Range
WS	21;5	8.57	9;10-34;9
DS	23;6	8.48	9;11- 35;3

Procedure

The British Ability Scales were administered to each participant. These scales allow for assessment of “carefully defined specific abilities” (Elliott, 1996, p.1). They also provide a measure of General Conceptual Ability (GCA), an IQ equivalent.

Participants were assessed using the six core scales: Recall of Designs, Word Definitions, Pattern Construction, Matrices, Verbal Similarities and Quantitative Reasoning. These can be used to construct a profile of verbal and visuospatial constructive abilities. Recall of digits forward was also included to measure phonological memory span, a reported strength in Williams Syndrome. The BAS provides standardised scores or T scores with a mean of 50, SD 10 and a range of 20-80 on each of the core scales.

The performance of each participant was compared to criteria developed by Mervis et al.. These criteria were chosen because they are claimed to characterise the classic cognitive profile of Williams Syndrome. In the Mervis et al. study, specific subscales from the DAS were assumed to reflect possible areas of strength and weakness. The strengths reported for language in WS were operationalised using two subtests: Verbal Similarities, which measures verbal knowledge and reasoning, and Word Definitions, a measure of expressive vocabulary. Scores from Recall of Designs which assesses memory for visuospatial relationships, and from Pattern Construction, a test of non-verbal reasoning and spatial visualisation, were used as measures of visuospatial constructive ability. Scores on these scales were predicted to be lower in the WS group than those for the language items.

3.3.2 Data Analysis

The same cognitive profile criteria were used as in the Mervis et al. study. In order to fit the WS pattern, participants' performance had to conform to the following:
(Standard scores, known as T scores on the BAS, were used for this analysis.)

Williams Syndrome Cognitive Profile (WSCP)

1. Pattern construction score < mean score for the six core scales

This means that visuospatial constructive functioning is lower than the mean level of functioning over all tests.

2. Pattern construction score < digit recall score

Here visuospatial constructive ability (a purported weakness in WS) should be lower than phonological short-term memory ability (a purported strength).

3. Pattern construction score < 20th centile

This reflects low visuospatial ability (expected to be well below the mean).

4. Score for either digit recall (short-term memory)

or word definitions (vocabulary)

or verbal similarities > 1st centile (T score > 29)

If an individual meets all 4 of these criteria, it means that either phonological short-term memory or language performance is relatively good. It is important to note that these criteria can be fulfilled, even if the participant's overall performance is poor or if a participant is unusually talented.

Degree of fit to the WSCP

If an individual met all four criteria, and therefore displayed the WSCP, further analysis of their performance was carried out. The strength or degree of their conformity to the pattern was measured, assigning scores by the following criteria:

a. Digit recall score > mean score for the six core scales 2 points

This is a strong test of the expected short-term memory advantage for WS. Here memory score has to be higher than the mean overall score (including language and non-language items).

b. Word definition score > pattern construction score 1 point

This reflects the relative strength of vocabulary over visuospatial abilities.

c. Similarities score > pattern construction score 1 point

This reflects the relative strength of verbal knowledge and verbal reasoning over visuospatial abilities.

3.3.3 Results

Only 5 of the 15 cases (data from 3 individuals) which were included in the WSCP analysis met the criteria for the “classic” WSCP. None of the DS cases conformed. For the WS group, data from 6 of the cases did not fit because all their subscale scores were at floor, i.e. a T of 20. This caused their mean T score to be 20 and meant that it could not be greater than any other measure. It was therefore impossible for any differences, necessary to meet the criteria, to be exhibited.

For the 5 cases that met the criteria, the degree of conformity to the WS profile was also assessed. A score of 4 points indicated a strong match to the profile. All cases that met the original criteria had the strongest possible match. This suggested that the WS individuals in this study who exhibited the “classic” WSCP did so very strongly.

The results of the analysis suggest that the cognitive profiles of the DS group are completely different from the classic WSCP. An additional analysis, which I devised and was not conducted by Mervis, giving one point for each of the main criteria met, gives the DS group a mean score of 1.3 out of a maximum 4. This type of analysis enables one to measure the degree to which the individual matches the criteria, giving information even when the full criteria are not reached.

A much smaller number of WS individuals than would be expected met the Mervis criteria, given her previous study. However, when each of the main criteria was assigned one point, as for the DS group, the WS mean score was 3.42, higher than the DS score. A one way Anova confirmed this, $F(1,23)=67.5$, $p<.01$. (See the footnote on page 43 for a discussion of the use of multiple data points from individual participants.) My WS sample may not conform absolutely to the Mervis characterisation of the WS profile, but they do come much closer to it than the DS group, as would be predicted. An alternative analysis of the cognitive profile may confirm the WS pattern and is described in the following section.

3.3.4 An alternative analysis

Many of the participants in the present study were at floor on several or all of the core subscales (with T scores of 20). It was obviously difficult for them to fulfil the criteria when all their scores, but not necessarily their underlying abilities, reached the same floor score. In order to overcome this, age equivalent scores were used to carry out the profile analysis. Within individuals, age equivalent scores varied across subtests so it was more likely that a pattern of strengths and weaknesses could be detected.

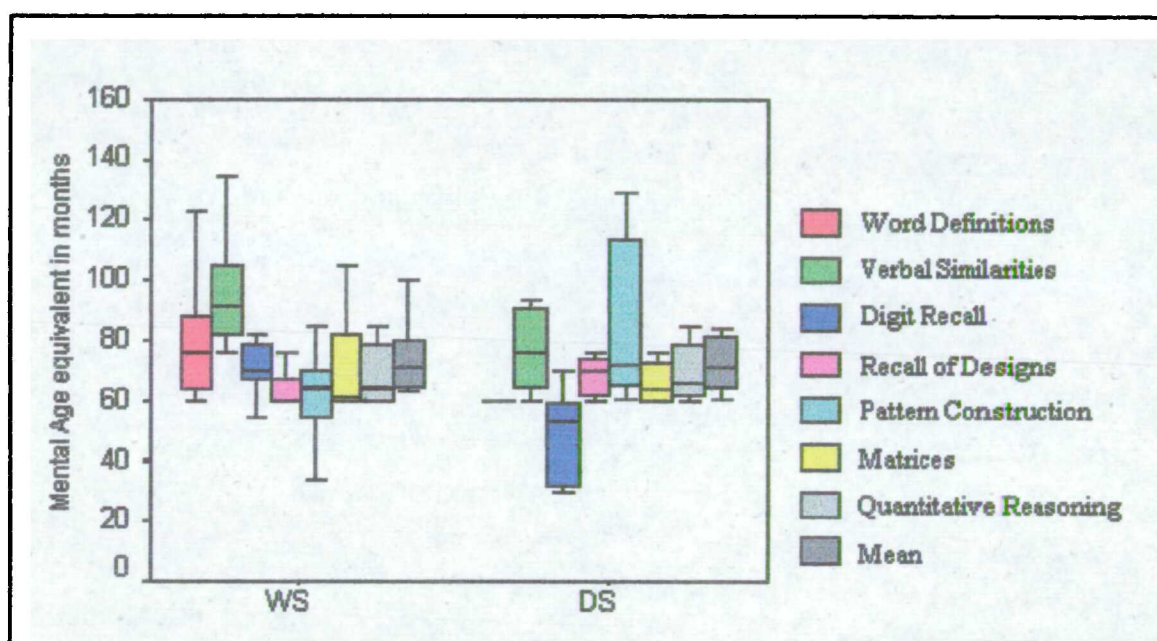
In the analysis, age equivalent scores were converted from years and months into months. Then, Mervis et al.'s criteria were used with the following adjustments. Where a T score was used in the original version, mental age was substituted. For criteria 3 and 4, where centiles were used, the mental age that corresponded to the centile for each individual was used.²

The analysis described above yielded clearer results than Mervis et al.'s procedure. In terms of age equivalent scores, a far better range was revealed. Patterns in the data thus became more apparent. Figure 3.1 illustrates the pattern of abilities in both the WS and DS groups. The box plot shows the distribution of performance in the sample.

² E.g. the 29th centile for a 17;0 year old for word definitions corresponds to raw scores of 116-118. If one takes 116 this corresponds to a mental age of 11;3. This must be done separately for each individual because centiles are age dependent.

The top of the box corresponds to performance at the 75th centile and the bottom of the box to the 25th centile. The horizontal line marks the 50th centile. Readers interested in data for each subtest from individual participants can find this tabulated in Appendix C.

Figure 3.1 Box Plot of Mental Age Equivalents for seven subtests of BAS-III (outliers excluded)



When mental age equivalents were used, 8 of the 15 profiles (data from 6 individuals) from the Williams Syndrome group conformed to Mervis' criteria. The removal of floor effects allowed the underlying profile to emerge. None of the profiles from the Down's Syndrome group conformed.

3.3.5 Discussion

Williams Syndrome

Just over half of the cases in the WS group conformed to the criteria, which reflected the WSCP. This was only possible when mental age equivalents were used, instead of T scores, to ensure a greater distribution of performance. The following section

examines a possible explanation for a lack of conformity to the WSCP for the whole WS group.

Which criterion caused the lack of conformity in the Williams Syndrome group?

Given the low level of conformity seen in the WS group, a criterion by criterion analysis was carried out to see whether one particular criterion was causing the WS group to fall short of the Mervis profile. The fourth criterion caused 6 of the WS cases (data from 4 individuals) to fail to conform to the WS profile. In order to meet this criterion, individuals had to perform better than $T = 29$ or equivalent mental age on either the digit span measure or one of two language measures. Many of the individuals in my WS sample did not have the language skills to meet this criterion. Here we should return to Mervis' sample. The mean IQ of the WS in that study was 59.32 (SD 10.74). In my study, the mean GCA, from those 5 individuals whose scores were valid for GCA analysis (up to age 17;11), was considerably lower, i.e., 41.3 (SD 5.74). If all individuals were included, resulting in an GCA estimate which was probably too high, the WS group GCA was even lower than that of the smaller subgroup and still lower than that for Mervis et al.'s sample. If language performance were spared and at GCA level, high scores would not be expected on a standardised test because of low overall ability. In addition, if better than average performance were expected, it would be even more difficult for the participants to meet the criterion.

In sum, the WS individuals in the present study do not all conform 100 percent to criteria, which have been claimed to be indicators of the classic WS profile, but do show a tendency towards this profile. It may be that the language expectations of the criteria are too high for many WS individuals, unless they are particularly high functioning. Poor visuospatial constructive ability appears to be a good indicator of the classic WS profile. When fit to the main criteria was considered, the WS group conformed to a much greater degree than the DS group.

Down's Syndrome

Constructing a Down's Syndrome Cognitive Profile (DSCP)

As discussed in the introduction, previous studies have revealed that, despite the suggestion of a uniformly flattened profile, a pattern of strengths and weaknesses exists for DS. This is almost the converse of WS and is less pronounced. Individuals with Down's Syndrome have difficulty with verbal memory and language tasks, but are better at visuospatial tasks (Jarrold et al., 1998). Given this pattern of abilities, one can try to match the DS participants to criteria that reflect a specific DSCP. This was done simply by reversing all of the Mervis criteria. For example, Block design performance was hypothesised to be better than language performance. Age equivalent scores were used, as these give rise to a better distribution of performance.

No DS subjects conformed fully to all the criteria, but this was not unexpected. It is known that the DS profile is more uniform than the WS profile. When fit to the DSCP is assessed, for both the DS and WS groups, with one point given for each of the main criteria fulfilled, the mean DS score of 2.77 (SD 0.44) is higher than that for the WS group (mean 0.53, SD 0.74). A one way Anova confirmed this, $F(1,23)=151.25, p<.01$. The reason for lack of conformity to the DS profile is considered in the section below.

Which criterion caused non-conformity in the Down's Syndrome group?

One particular criterion was particularly difficult for the DS group to fulfil. The participants were unable to score higher than the 20th centile on the block design task. This benchmark demands a high level of performance, one which does not reflect the mental ability of the participants in my study, whose mean GCA is only 41.3 (SD 3.39). If this stringent criterion is removed then 4 of the 6 participants conform to the pattern. In general, the language skills of DS participants, e.g. verbal similarities and word definitions, are much lower than their visuospatial constructive abilities, as made clear in Figure 3.1. This is the opposite of the WS pattern, as predicted.

3.3.6 Conclusions

It appears that a classic profile of WS, at least as operationalised by Mervis and her colleagues, is not seen in all individuals with this condition. That said, if results are analysed in a less stringent fashion, investigating the degree of fit instead of absolute conformity, the WS group do conform to a much greater degree than those individuals with DS. In my view, degree of fit is more useful than absolute fit for a number of reasons. A less stringent approach allows for variation within the phenotype. We know that the performance of individuals with WS is very widely distributed, with IQs ranging from 40 to 70. In addition, the degree to which the profile is uneven varies across the group. If the degree of fit is assessed, then individuals with WS can be placed along a continuum, from a very strong fit to the classic profile to a rather less well-defined WS cognitive profile. Degree of fit also allows an individual to fit to the profile despite falling down on a particular criterion. Several individuals in my sample were unable to meet the high language requirements of the Mervis criteria, but when their degree of fit was measured, it could be 3 out of the maximum 4. In addition to using the inverse of the WSCP, an attempt was made to formulate a specific cognitive profile for DS. However, this did not yield a profile because of the small amount of data available from which to extract a consistent pattern. The individuals exhibited many different patterns, but did conform to the inverse of the WSCP, which was of course not originally formed from their data.

It is clear that the WSCP is distinctive. None of the DS group conformed to the Mervis criteria and their degree of fit was also very low. Therefore, the WS profile is not just a product of retardation but is likely to be syndrome specific. An attempt to fit the DS participants to a DSCP, formed from the inverse of the WS profile, was partially successful. No DS subject conformed to the criteria absolutely, but the degree of fit to this inverse of the WSCP was high. It seems that different syndromes do give rise to qualitatively different cognitive profiles. The profiles of infants with WS and DS differ to a lesser degree than those exhibited in middle childhood and adulthood. Therefore, the different developmental trajectories followed by these syndromes, and not solely innate mechanisms, must be responsible for the cognitive profiles present in the steady state.

3.3.7 Suggestions for improvements and further research

This chapter considered variations on one method for characterising the WS cognitive profile, that used by Mervis and her colleagues (1999). I have suggested that measurement of degree of fit to the profile may be as useful, or more useful than absolute conformity of an individual. It would be interesting to reanalyse Mervis' own data using this method.

There may also be other ways of characterising the WS profile. A survey of all attempts to show good language ability in the face of poor visuospatial constructive ability may highlight tasks that could be used to construct a list of criteria for the WS profile. For example, within the BAS, recall of designs could be included to strengthen the assessment of visuospatial ability. In my sample, in 13 of the 15 WS cases (data from 6 individuals) performance was below their overall average on this measure.

A first attempt was made to create a DS profile, using the inverse of the Mervis criteria. Further work could refine these criteria to allow for the flatter cognitive profile in DS and for the overly high expectation of performance on the block design task. It might also have been interesting to compare different forms of Down's Syndrome against the DSCP. Perhaps individuals with different genetic profiles, e.g., mosaicism or translocations, would have slightly differing cognitive profiles.

3.4 Chapter Summary

This chapter provides a characterisation of the Williams Syndrome Cognitive Profile, to which most of the WS sample conform to some degree. The DS sample does not conform to this profile and an attempt was made to provide criteria for a Down's Syndrome Cognitive Profile. This group displayed a less well-defined profile, as is sometimes reported in the literature. The data suggest that WS and DS groups do exhibit different cognitive profiles in adulthood, a pattern that is not yet present in the infant phenotype.

Now that the general cognitive profiles of infants and adults with WS and DS have been considered, the following chapters will examine specific aspects of cognition.

Specific profile: Language development in infants with Williams Syndrome and Down's Syndrome: Data from a questionnaire and two experimental studies

Introduction

This chapter examines two specific aspects of language development in infants with Williams Syndrome and Down's Syndrome compared with typical development. The first two studies investigate the extent of single word production and comprehension in each group and the third is an attempt to examine one of the first stages of syntactic understanding, word order. This is the first time that such a systematic characterisation of the very early stages of WS and DS language development has been carried out. It is an important step for a number of reasons. The literature on adult language ability in WS and DS points to striking differences between the two groups. Adults with WS perform well on language tasks in comparison with their non-verbal abilities, whereas those with DS have difficulty with language throughout life (e.g., Bellugi, Marks, Bihle & Sabo, 1988; Chapman et al., 1991, 1998; Mervis, Morris, Bertrand & Robinson, 1999). Although it is known that WS language is not intact, with problems in several micro domains, it remains clear that differences between the DS and WS groups are very marked. But do these differences already exist in infancy? This chapter sets out to investigate this, by looking at infant precursors to later language ability. This is an attempt to examine whether the cross-syndrome differences in language abilities seen in the steady state are already present

in infancy or whether infant performance is the same in DS and WS. The latter would suggest that the differences later in life are a product of different developmental processes, affecting learning and brain organisation. An investigation of early language may provide a window onto the developmental trajectory that each group takes.

The studies presented here will also address the character of early language development in WS and DS. In the atypical development literature, there has long been a discussion as to whether atypical development should be characterised as delayed, merely slower than normal but on the same track, or whether such development is actually deviant. If the latter were true, then language development in atypical groups would unfold in a different manner.

The existing language literature from DS, WS and normal developmental research will be reviewed in part one of this chapter, and the results of the present study (part 2) will be examined in the light of this previous research. Particular emphasis will be put on language comprehension, given the age of the individuals involved in my research. Many of the infants who took part were only just beginning to produce any language at all. This literature review will be followed by specific reviews in the introduction to each of the three empirical studies.

Part One: Literature Review

This review will be presented in three sections. The first will be an examination of the literature concerning language development in typically developing infants. In particular, I examine results that highlight those areas in which infants with WS are known to exhibit unusual patterns of development. Data from studies of normal children employing similar methods to those in the empirical section of this chapter are also included, in order to provide a benchmark for the results from the atypical infants. In addition, the results of studies using electrophysiological measures of brain activity in normal infants are presented because this technique has great potential to reveal subtle abnormalities in cognitive processing. The second and third sections describe studies on infants with DS and WS respectively. The WS section includes a brief overview of some studies examining brain activity because they point to further atypical language development in this group.

4.1 Normal infants: lexical development

There are several universal links between language and cognition which are thought to hold for both typically and atypically developing children. Some of these will be discussed below and their presence in clinical groups will be considered later. It is expected that the pattern of abilities in older typically developing infants and children will also be apparent in younger normal infants when the precursors of lexical ability are studied.

4.1.1 Making sense of linguistic and non-linguistic input

In order to build a lexicon, the infant has to be able to decode both linguistic input and the non-linguistic cues referring to objects or actions in the environment. To do this the child must puzzle out what the utterances he hears are referring to, making use of any clues available, such as pointing or movement of the specific object into their attentional space. Certain fundamental lexical principles become available as the child develops to aid in this complex task of disambiguating meaning. While some

authors argue that the principles are innately specified (Hall & Waxman, 1993; Markman, 1994), others have shown that they follow an experience-dependent course (Mervis & Bertrand, 1993; Gathercole et al., 1995, Golinkoff et al., 1994). The following sections will outline these principles in both cognitive and linguistic forms, as their use is apparent in both.

Pointing and language

Normal children begin to point to objects in the environment before they can label them. This is known as referential or declarative pointing and can be thought of as a substitute for the ability to label the object. Pointing out an object to a caregiver enables the child to focus the adult's attention on it and often generates labelling from the adult. In normal infants, understanding of the referential pointing of others, used to establish joint attention, precedes the child's ability to point. At 9 months, normal infants will look at the object being pointed out, and by 10 months they will point out objects themselves (Murphy & Messer, 1977). Pointing enables a normal child to build up a store of labels before being able to ask for them orally. Even before this, infants exert some influence over the linguistic input they receive. At around 7-9 months, they begin to babble rhythmically. This rhythmic quality makes their output sound more like speech. This in turn causes the adults to provide a greater amount of linguistic input because they believe that the child is trying to express something understandable.

Lexical constraints

Although pointing focuses the attention of both child and adult, it goes only a little way towards disambiguating the referent of a label. For example, an adult may point to a cat and say "cat", but how does the child know to what this label refers? It could refer to a furry four-legged animal, to that particular instance of cat, or could refer to a part of the cat (e.g. fur or collar or even its colour). Infants and toddlers use several lexical principles to constrain this potential ambiguity of reference. Firstly, they assume that a novel word refers to a whole object. So if a toddler is presented with a teapot, he would assume that a label given to it referred to the whole object and not to

just the spout. However, using the mutual exclusivity constraint the toddler would also assume that if he already knew that the name for the object was "teapot", then the novel word must refer to a part, e.g., the spout, because whole objects are normally given only one name. These assumptions can be investigated before the emergence of productive language by examining the way in which children treat the objects around them, for example the way in which they sort objects into groups (Mervis & Long, 1987).

Apart from the mutual exclusivity principle and whole object constraint, another, the Novel Name-Nameless Category (N3C) principle, can be used by children to reduce the number of possible referents a new label could have (Mervis & Bertrand, 1994). Children realise that all objects should have a name; a realisation that can be assessed using the fast mapping paradigm (Carey & Bartlett, 1978). If a child is presented with three objects, one of which is unfamiliar, she will map a novel name to that object. This fast mapping ability coincides with the emergence of spontaneous exhaustive sorting, reliant on the child's assumption that all objects belong to a particular category. It also coincides with the onset of the vocabulary spurt, the acquisition of new words having been made more efficient by the N3C principle. In both longitudinal (Gopnik & Meltzoff, 1987; Mervis & Bertrand, 1994) and cross sectional studies (Gopnik & Meltzoff, 1992), it was found that those 16-20 month infants who could sort a mixture of objects exhaustively into two categories were also those who used the N3C principle, and those who did not sort spontaneously and exhaustively had not yet begun to fast map. As mentioned above, the vocabulary spurt is closely related temporally to both of these abilities. This link can be charted by looking at the progressive increase in the number of words produced by toddlers over time. There are various estimates of the amount of words that need to be acquired in a given period of time to constitute a spurt, but a reasonable definition would be that the spurt occurs when the child suddenly starts to acquire as many as 10 new words in a two week period (Mervis & Bertrand, 1994). In a normal infant, the spurt occurs when the child has a lexicon of around 75-150 words. The following section examines the kind of words that appear in early productive vocabularies of typically developing infants.

First words

First words tend to be used for basic level categories. This is thought to be the most fundamental level of the category structure used by adults. Basic level categories share similar features and functions which are easily discriminated from those of members of other categories. Ball is an example of a basic level category, whereas football (subordinate) or sports equipment (superordinate) is not. All balls share common attributes and have broadly the same function. However, they are nothing like cars, another basic category. In the case of cars, Volkswagen is a subordinate category and vehicle is the superordinate. The first words children produce are almost always basic level category labels (Mervis, 1983). It should be noted, however, that the meaning of a basic level category for a child might not be the same as that used by an adult. Children often focus on attributes that are not deemed important by adults. The opposite may also obtain. They sometimes ignore a feature that is important for distinguishing between two categories for an adult.

4.1.2 Mental age and vocabulary acquisition

Now that the type of constraints necessary for vocabulary acquisition have been considered, it is important to examine the links between language development and general cognitive ability. It may be that links found in typically developing infants between certain stages of cognitive and language development are not found in neurodevelopmental disorders which follow a different developmental trajectory.

Mervis examined the size of the lexicon in relation to mental ability for both typically and atypically developing toddlers on two measures: The Bayley Scales of Infant Development and Piagetian object permanence tasks. The vocabulary spurt usually occurs when the infant has reached Stage VI in object permanence tasks and has a MA of between 18 and 20 months on the Bayley Scales of Infant Development (Cardoso-Martins & Mervis, 1985). In a longitudinal study, Mervis showed that typically developing infants had a mean MA of 13.8 months when referential comprehension was first documented and an MA of 19.5 months when production was first noted. It is important to note that Mervis is very clear that production in this

case must be referential, i.e., in response to the demand for a label for an object from an experimenter and not merely a context-bound (Harris, Yeeles, Chasin & Oakley, 1995). It was not sufficient for a child to understand a label only in a specific context. For example, the child was required to link the label “cat” to any instance of a cat and not only a particular cat. This might explain why the mental age levels she reports seem rather high, in the face of data from questionnaire studies.

4.1.3 Examining the receptive lexicon: behavioural techniques

The studies reported in Part Two of this chapter examine language comprehension rather than production, because of the age of the participants tested. The following section presents findings from studies which use two different methods to assess language comprehension. The results from these studies with typically developing infants can be used as a benchmark when considering the performance of infants with WS and DS, on which tests of this kind have not been conducted up until now.

As well as measuring the amount of vocabulary produced, it is also possible to assess the receptive language abilities of an infant. This ability precedes the ability to produce words. Huttenlocher (1974) suggests that in order to produce a word, the child must recognise the referent, recall its label, and articulate the sound of the word. By contrast, to understand a word, the child must recall the referent and only recognise the label, a task which is less demanding cognitively. In general, there is indeed a considerable lag between comprehension and production. On average, 16 month olds in the USA have a receptive vocabulary of 200 words but are able to produce only 27, as reported on the CDI (Fenson et al., 1994)

Receptive vocabulary is commonly studied using a preferential looking or listening technique. Using the listening technique, it has been shown that at 8-12 months an infant will prefer to listen to words he recognises in nursery rhymes rather than substituted nonsense syllables (Glenn & Cunningham, 1982). Researchers have shown that at 12 months, infants will listen preferentially to lists of familiar words over rare unknown words even when no other non-linguistic support is given (Hallé et

al., 1994). In one experiment, lists were presented to 11 and 12-month-old infants from either their left or right side and children turned more often to the side from which the familiar words were coming. To rule out the possibility that infants were basing their preference on phonological complexity, preferring familiar words because they were less complex, further experiments were carried out in which rare and familiar stimuli were matched on complexity. The same familiarity effect was found. In the 11-month-olds, this preference was not as strong as in the 12-month-olds, suggesting that comprehension is just emerging at this time in development.

Preferential looking techniques can be used with older infants to investigate the links they form between spoken words and the objects, whether real or pictures, to which they refer. In this paradigm, the child is presented with pictures of two objects and one of the objects is named. It is expected that the child will look significantly longer at the named object if he understands the label given. This preferential looking pattern was found for nouns and verbs (Golinkoff et al., 1987) in 13 month olds, but not at 11 months (Thomas et al., 1989, Behrend, 1987). It has since been extended to use real objects and to test the acquisition of lexical constraints (Hollich et al., in press).

4.1.4 Examining the receptive lexicon: brain imaging techniques

In addition to the use of behavioural techniques such as those described above, it is now possible to assess comprehension using ERPs (event related potentials). By measuring the electrical activity of the brain with sensors attached to the scalp, one can map the neural correlates of overt behaviour. This approach may be particularly fruitful when working with atypical groups as unusual electrical activity may be found to underlie seemingly normal behaviour but atypical cognitive processes. Already, changes in the brain over typical development have been highlighted by using such techniques. For example, it has been found that differential brain responses occur in children when they are presented with words they know and words they do not. For 14 and 16-month-old infants, data indicated that their responses differed on three counts: their knowledge of the word, presentation order, and chronological age

(Molfese, 1989, 1990). These developmental findings may have relevance in the atypical case because differences in the responses of DS and WS may reflect immature or completely different processing.

Differences in brain organisation in relation to language competency have also been measured. This has been done both with normal adults (Neville, 1991a,b; Neville, Kutas & Schmidt, 1982) and with children with Specific Language Impairment. The latter group is of interest here because the results suggest that language difficulties seen in this population are linked with atypical patterns of brain activity (Neville, Coffey, Holcomb & Tallal, 1993). In general, Neville et al.'s data indicate that the electrical activity associated with language processing was more localised in the left hemisphere in those individuals with greater language competency.

A study of normal 20 month old infants with differing sizes of productive vocabulary and receptive vocabulary shows that the changes in brain activity reported by Neville et al. (1993) are indeed due to language competence and not merely to maturation. Those infants with larger vocabularies showed both within and across hemisphere differences in activation when presented with known or unknown words auditorily, whereas those with low vocabularies showed symmetrical activity in both hemispheres (Mills, Coffey-Corina & Neville, 1993). These findings are of interest for atypical development. One can ask whether atypical infants will show this type of progressive cortical specialisation for language as normal infants do, and whether this is more apparent in those with greater language competency.

4.1.5 Language comprehension and production: a dissociation of abilities?

Now that techniques for assessing receptive language have been discussed, I will turn to the relationship between receptive and productive language. There has been some debate over the emergence of production and comprehension in language development. Some investigators provide evidence that production precedes

comprehension (Chapman, 1977), whereas others have data showing that comprehension begins first (Bates, Bretherton & Snyder, 1988). Recent reports support the latter view, although there is a great deal of individual variation in the data (Harris et al., 1995). Using a looking time measure, Golinkoff, Hirsh-Pasek, Cauley and Gordon (1987) have shown that infants are able to understand single nouns, independent of context, very early in development before 14 months. However, infants begin to produce their first object labels, independent of context, at around 18 months (Cardoso-Martins, Mervis & Mervis, 1985). The question that will be addressed in the WS and DS sections is whether a similar pathway is followed by atypically developing infants. Does production emerge after comprehension as in normal development or is the developmental course different?

4.1.6 Lexical and syntactic comprehension: a dissociation of abilities?

It is likely, then, that in the normal case comprehension is in advance of production, but is there also a difference between competency in lexical and syntactic comprehension? Looking time paradigms provide evidence that noun comprehension is in advance of verb comprehension. Infants also begin to understand word order later than single words. (Golinkoff et al., 1987, Hirsh-Pasek & Golinkoff, 1996). This is an expected finding. The infant must understand the components of a sentence, i.e., what the nouns and verbs mean, before we can investigate whether he understands the role of, say, word order in syntax. If presented with a sentence such as, “The duck is pushing the cat” the child has to attend to both nouns around the verb, and distinguish the two, in order to decode the word order. Data from looking paradigms show that normally developing children begin to understand words at around 13 months (Thomas, Campos, Shucard, Ransay Shucard, 1981) and start understanding aspects of grammar such as word order at 17 months (Hirsh-Pasek & Golinkoff, 1996). Thus, there is a lag between comprehension of words and the comprehension of basic syntax. While the presence of an asymmetry in the development of these two aspects of language is recognised, it is important to note that it is the degree of delay between lexical and syntactic understanding that is interesting. To this end, the pattern of

development seen in atypical development will be considered in relation to the normal pattern. In atypical development, it is likely that there is a larger discrepancy between the onset of these abilities than that seen in typical development

We turn now to language in atypical development for the two syndromes under study: Williams Syndrome and Down's Syndrome, which display different patterns of language competency in later development. First, Down's Syndrome will be considered.

4.2 Down's Syndrome: lexical development

In general, it has been found that when the steady state is reached, language skills lag behind other cognitive skills in those with DS, a profile inverse to that found in WS (e.g., Mervis et al. 1999, Chapman, 1995). The following section is a review of the relationship between cognitive and language abilities in the early stages of DS language development. In particular, it addresses the question of delayed versus deviant development.

4.2.1 Making sense of linguistic and non-linguistic input

Rhythmic babbling and hand clapping appear at roughly the same time in normally developing children. Mervis et al. (1997) demonstrated that this synchrony was also true of infants with DS. As mentioned with regard to typical development, rhythmic babbling means that infants with DS sound as if they were producing speech-like utterances and therefore receive more input from the adults around them. This, in turn, should aid the development of the infant's own language production.

Pointing and language

Referential pointing to objects also occurs in infants with DS, with production of the first referential object words coming soon after pointing emerges (Mervis et al., 1997).

Lexical constraints

Infants with DS abide by the same lexical constraints as typically developing infants. For example, they assume that new labels refer to whole objects in the environment (Mervis & Long, 1987), as do their parents when talking to them and as do normal infants (Shipley, Kuhn & Madden, 1983). In addition, Mervis and Bertrand (1995) reported that in Down's Syndrome the link between the vocabulary spurt, fast mapping and exhaustive sorting is the same as that found in normally developing children. This indicates the presence of the N3C principle, which begins to be used when the infant is aware that all objects have a name. In a cross sectional study examining these links, 29-42 month old toddlers with DS showed one of two patterns: either they demonstrated exhaustive sorting of objects and had already gone through the vocabulary spurt, or they exhibited neither (Mervis & Bertrand, 1994). The same was true of fast mapping and the vocabulary spurt (Mervis & Bertrand, 1995). These various findings suggest that the relationship between early language development and other cognitive skills, or between different language skills, is similar in DS to that present in normal development, albeit later in DS.

First words

The lexicon used by children with DS is organised in a way similar to that found in their typically developing counterparts. 83% of the first 50 words produced by children with DS are the same as those found in normal children (Gillham, 1979). Both groups of children build up a bank of basic-level object words, and include the same types of objects in these categories. In a longitudinal study charting the development of categories in 6 DS subjects aged 17-19 months and 6 mental-age matched typically developing infants, aged 9 months (see Mervis, 1990), both normally developing children and those with DS showed a preponderance of basic-level category labels, such as ball and car, in their lexicons. They included items under these category labels that were appropriate for children but not adults, due to over- or under-generalisation of certain form-function attributes. So, for example, children would call a spherical candle a ball (ignoring the wick) or fail to call a

cement truck a truck because it did not fit the criteria the child used. This behaviour is also characteristic of typically developing children.

One particular difference that emerged between the DS and normal environments was the way in which parents labelled objects for their infants. The role of parental input into language acquisition must not be underestimated (e.g., Clibbens, 1993; Harris, 1992). 67% of mothers of normal infants, but only 31% of the mothers with DS infants, used the basic level name for an object on the day the child first understood it. The mothers of DS infants, by contrast, more frequently used the precise object name (e.g., lion) than the more generic basic level term (e.g., cat) (Cardoso-Martins, Mervis & Mervis, 1985).

4.2.2 Mental age and vocabulary acquisition

Although receptive vocabulary may be at the same level as found in normal MA-matched subjects, productive vocabulary in DS is often well below their mental age. Despite the finding that exhaustive sorting, fast mapping and the vocabulary spurt all coincide in DS as in normal development, toddlers with DS are older than normally developing children when the vocabulary spurt begins. This is because of their general developmental delay, and the slower rate at which children with DS acquire new words. In one study, normal children with a MA of 21 months had a mean productive vocabulary of 118 words, whereas 36 month old participants with DS at the same MA level could produce only 18.5 words (Strominger et al., 1984). This difference is probably accentuated by the timing of the vocabulary spurt in DS children. At 18 months, normal children have a vocabulary of approximately 22 words, not much above that of the DS subjects. However, the development of vocabulary of normal subjects is non-linear and shows a significant increase between 18 and 21 months. In support of this difference between the early and later stages of language development, a recent study of Italian children with DS suggests that this discrepancy in production ability between DS and MA-matched groups may not be present at an early stage of vocabulary development, that is before an MA of 17 months (Caselli, Vicari, Longobardi, Lami, Pizzoli & Stelli, 1998).

Despite the majority of children with DS following the normal developmental pathway, in some children the vocabulary spurt can come up to two years after both the ability to fast map and to sort objects into categories (Mervis and Betrand, 1997). This suggests that there is a wide within-syndrome variation in the size of the lexicon and its speed of acquisition. DS infants begin the vocabulary spurt when they have a considerably larger lexicon than typically developing infants.

There are several reasons for the difference between DS and normal vocabulary despite the fact that both groups follow a similar pathway in the development of lexical principles. These will be considered in the following section.

These findings suggest that in many respects children with Down's Syndrome acquire lexical principles in the same order as normal children, although they show considerable delay in vocabulary learning, in terms of age of acquisition. This is particularly true of lexical production.

The following sections survey the differences in development within language. The first discusses the discrepancy between comprehension and production abilities and the second, the discrepancy between the lexicon and syntax.

4.2.3 Language comprehension and production: a dissociation of abilities?

Language development in DS is normally characterised as delayed (e.g., Mahoney, Glover and Finger, 1981; Wisniewski et al., 1988; Singer-Harris, Bellugi, Bates, Jones and Rossen, 1997). Despite this delay, within language there are strengths and weaknesses apparent in this group. Studies have shown that children with DS have much more difficulty producing language than understanding it (Chapman, 1995; Cicchetti & Beeghley, 1990; Miller, 1987, 1988; Rosin, Swift, Bless & Vetter, 1988). The discrepancy in abilities is apparent from about 17 months (Miller 1988, 1992 ab; Stella, Lami, Caselli, Casadio, Pizzoli, 1993), when production begins to lag behind comprehension to a great extent, even in relation to mental age. Several studies

suggest that infants and young children with DS have comprehension abilities at the level of their mental age (Cardoso-Martins, Mervis & Mervis, 1985; Carr, 1988; Rosin, Bless, Swift & Vetter, 1988; Chapman, Schwartz & Kay-Raining Bird, 1991). These infants begin to understand their first words at the same time as their mental age-matched peers. Infants with DS whose MAs are around 13-21 months have the same size receptive vocabulary as 13-21 month old typically developing infants (Cardoso-Martins, Mervis and Mervis, 1985). It should be noted, however, that some researchers report that individuals with DS lag behind even in comprehension (Miller, 1988).

In contrast to comprehension ability, individuals with DS have considerably more problems with production than would be predicted from their MA (Strominger et al., 1984; Miller 1988, and many others). The extent of these difficulties may not be as apparent in the very early stages of language acquisition, however. Cardoso-Martins and her colleagues (1985) report that the onset of referential production occurred at a mental age of 19.5 months in their DS group and at 18.9 months in their typically developing group. Despite this, a lag in vocabulary size was apparent at an MA of around 21 months in the DS group, suggesting a slower rate of acquisition. In addition, if infants are matched with typically developing 13 month old counterparts on verbal comprehension levels, then production levels are the same in both groups (Caselli et al., 1998). This highlights the importance of the age at which studies are conducted. A normal 13 month old would have very little productive language, so differences between groups would be less obvious. However, 5 or 6 months later, at the time of the vocabulary explosion, the gap would be much greater.

Several possible reasons for this discrepancy between production and comprehension have been put forward. These include hearing problems, motor difficulties and differences in input received by an infant with DS. Hearing problems would lead to a decreased ability to store a good representation of the target to be produced. Motor difficulties lead to problems in planning and executing the articulatory programs necessary to co-ordinate the mechanisms used to produce speech (Miller, 1992a). As mentioned above in the section on making sense of linguistic and non-linguistic input,

the linguistic input that infants with DS receive differs from that available to their typically developing counterparts. In addition to differing in the type of categories used, parental speech to infants with DS is directive and less semantically contingent for the child than that experienced by normally developing children. These features of parental style have been found to correlate negatively with vocabulary growth (Cardoso-Martins and Mervis, 1985). Some of these differences, such as hearing loss, are not syndrome specific although others may be more so. For example, verbal memory is particularly poor in individuals with DS (Buckley, 1997; Marcell & Armstrong, 1982).

4.2.4 Lexical and syntactic comprehension: a dissociation of abilities?

As well as the decalage between productive and receptive vocabulary, individuals with DS have more difficulty with morphology and syntax than with single words. Their syntactic comprehension ability lags behind that of MA- matched children (Miller, 1988; Rosin et al., 1988; Chapman et al., 1991). Chapman and her colleagues (1991) found no differences between DS children's scores on the Peabody Picture Vocabulary Test, nor on the TACL-R (a test of syntactic understanding) when compared to MA-matched children. However, the difference between these abilities within the DS group was more marked. On the TACL-R test, the individuals with DS (aged 5–20 years) had much more difficulty with the grammatical morphemes items and the elaborated sentences and reversibles items than with word classes and relations. This adds further weight to the hypothesis that lexical abilities are relatively spared in DS. As with the production /comprehension difference, at the very earliest stages of syntactic development the difference may be less apparent, with differences increasing over time (Fowler, 1990).

Why then is there such a discrepancy? Obviously, the size of the lexicon is important for development of productive syntax (Bates, Bretherton & Snyder, 1988). So even despite relatively spared lexical skills, individuals with DS may not have the resources available to them to begin the path to syntax. However, it is likely that this

would not affect comprehension to such an extent because the individual would not have to construct sentences themselves. Another possible cause of the discrepancy might be that individuals with DS have enhanced lexical ability that could lead to an apparent but relative syntactic disadvantage. Because they are older, individuals with DS have had much more exposure to vocabulary than their MA-matches and this could lead to a lexical advantage despite developmental delay. However, despite this explanation being intuitively plausible, it has been found that there was no advantage for DS children or other children with mental retardation over MA-matches on a test of receptive vocabulary (Rosin et al., 1988). With this explanation ruled out, there remain a number of further causes for the syntactic difficulties reported. The verbal short-term memory deficit found in individuals with DS might be a plausible explanation (Marcell & Armstrong, 1982; Marcell & Weeks, 1988). If the children with DS form poor representations of verbal input, then longer language samples will be more seriously affected than single words. This could lead to a particular problem with syntax because several words must be held in memory in order to construct a sentence or decode its meaning. However, Chapman et al. (1991) found that lexical comprehension could be predicted effectively (84% of variance) using CA, MA and hearing status alone.

Hearing loss is another possible contributor to problems with syntax and morphology. Noun or verb inflections are often very subtle and could easily be missed if auditory acuity is low. Chapman et al. (1991) did find that hearing contributed to comprehension problems. However, its effects did not differentiate between inflectional morphology difficulties and the more general understanding of longer items, as would be expected. Perhaps, individuals with DS focus on different aspects of the language when processing it. Bridges and Smith (1984) and Fowler (1990) suggest that individuals with DS place far more emphasis on semantics than on the syntactic structure of the language when decoding word order. This reliance on semantics may cause difficulty when it is not available to provide support for comprehension. Nevertheless, many of the differences, seen in performance are best explained by mental age- the individual's level of non-verbal cognitive ability (Miller, Chapman & Bedrosian, 1978; Abbeduto, Furman & Davies, 1989).

4.2.5 Summary

This review highlights several important points. There are possible changes in the relationship between lexical and syntactic understanding as development progresses. In order to capture these changes, it is important to characterise development from the very early stages. It is also clear that in making a survey of language ability, it is crucial to look at a number of areas within this larger domain. Despite the fact that DS language is delayed in terms of chronological age, the review has shown that certain areas of language are more affected than others. It appears that while individuals with DS have trouble with lexical production, their comprehension levels are much higher. In addition, they have particular difficulties with syntax, whereas their lexical abilities are at mental age level. This fact highlights the importance of investigation within domain differences as well as those between domains. The differences in which areas are affected across syndromes may be a key to understanding the mechanisms of typical development. If particular cognitive impairments are present in a syndrome in conjunction with a particular deficit in language, then it might be possible to examine the link between the two, and to investigate whether the link is important in typical development.

4.3 Williams Syndrome: lexical development

The pattern seen in the cognitive profile of children with Williams Syndrome is the inverse of that found in those with Down's Syndrome. Mervis et al. (1997) found that when Bayley scores were analysed for WS in terms of language and non-language items, there were a higher number of verbal items correct. There is also evidence to suggest that relative to non-verbal IQ, adults with WS have large and varied vocabularies (Bellugi et al., 1988; Rossen et al., 1996; Stevens & Karmiloff-Smith, 1997). However, the development of the lexicon may well be delayed and follow a course different from that found in normally developing or DS children.

4.3.1 Making sense of linguistic and non-linguistic input

Despite this relative strength in the verbal domain, children with WS use strategies to acquire their lexicon that turn out to be different from both normal and DS children. Whereas the DS children showed evidence of all five of the links between cognition and language found in normal children (rhythmic babbling, referential pointing, child basic categories, spontaneous exhaustive sorting with fast mapping, and with the vocabulary spurt), WS children only display simultaneity of development in three of these. Rhythmic babbling and its cognitive equivalent, hand clapping, appear at approximately the same time (Mervis et al., 1997). WS children also acquire basic category labels before superordinate and subordinate levels, as normal children do. This is seen both in referential production and in the way in which children play with toys belonging to the same child basic category (Johnson et al., 1994). Fast mapping and exhaustive sorting occur at roughly the same time in the child, suggesting that individuals with WS, like those with DS and normal children, realise that objects belong to categories and that each of these categories has a different label.

More interesting though, from the viewpoint of divergent developmental pathways, are the ways in which WS children do not follow the route taken by normally developing and other mentally retarded groups. WS children differ on two counts, one of which occurs at the very beginning of development of lexical production in normal children and the other which occurs later.

Pointing and language

Unlike both normal and DS infants, those with WS do not point to objects referentially before they produce their names. In fact, pointing follows on average only 6 months later (Mervis et al. 1997). This lack of pointing before naming is not due simply to a physical inability to point, because unlike normal 9-10 month olds (Murphy and Messer, 1977) WS children also do not respond to the pointing of others. Referential pointing is a means of focusing joint attention on a particular object so that, in the case of lexical acquisition, it can either be labelled for a child who does not know its name or so that the child can label it (Dunham et al., 1993).

Children with WS achieve this joint attention, but by other means, such as being provided with names for objects their caregivers see them attending to

Lexical constraints

The second difference in the pathway taken by children with WS is in the use of lexical constraints and in the timing of their vocabulary spurt and its links to spontaneous exhaustive sorting and fast mapping. In WS the vocabulary spurt often considerably precedes the ability to sort objects into categories. A discrepancy of 6 - 12 months was found from the onset of the vocabulary spurt to the emergence of exhaustive sorting. Some children had a vocabulary of 500 words plus before they began spontaneous sorting of objects into categories (Mervis et al., 1997). Despite this difference, fast mapping was still apparent in the WS group when the children began sorting into categories. These results suggest that individuals with WS might be focusing less on semantics in their word learning than their typically developing and DS counterparts. It appears that they are acquiring labels before acquiring the notion of object kinds or categories on which to map them. It might be that the WS group is learning the phonological sequences of labels but has only a shallow referential understanding of them (Karmiloff-Smith, Brown, Grice & Paterson, submitted).

In a study of older WS children and adults, differences were also found in the use of other lexical constraints such as the whole object and the taxonomic constraint. Unlike normal three-year-olds, subjects with WS used a whole object label equally for a part of a familiar or unfamiliar object. Normal children are more likely to use the whole label for an object with which they aren't familiar and to assume a label corresponds to a part if they are familiar with the whole object label (Stevens & Karmiloff-Smith 1997). In other words, the WS group tends not to obey the whole object constraint. WS individuals also made little use of the taxonomic constraint. When presented with a novel object that was given a novel name, and asked to give another object with the same label, WS subjects did not show a bias towards objects from the same taxonomic category. Again this constraint is in place in normal three year olds (see also Golinkoff et al., 1994). These particular constraints seem to be late

appearing and, given the delay in language acquisition in WS, may be never be acquired by these individuals.

Strategies for word learning in Williams Syndrome

As described above individuals with WS do not use the same constraints for word learning as typically developing infants or infants with DS. How, then, do those with WS support their word learning? It is possible that verbal short-term memory plays a large role in this process (Mervis et al. 1997; Grant, Karmiloff-Smith, Gathercole, Paterson, Davies, Howlin & Udwin, 1997). Indeed WS children have good short-term memory skills that are present from around two and a half years. Digit spans are sometimes found to be within the normal range (Finegan et al., 1995). This ability may enable the children to form representations of the words they hear given as labels by adults and then to repeat them when they see the appropriate object themselves. They may also pay particular attention to inputs of this type above all others because they are particularly well equipped to deal with them. However, the phonological memory abilities of children and adults with WS are impaired. Despite MAs of well over 4 years, these individuals rely on similar processes to normal four year olds, and do not even reach the level of five year olds who begin to rely on other properties of the lexicon to support word learning (Grant et al., 1997). It is important to note that digit span, in which people with WS show some proficiency, is based on monosyllabic words. By contrast, tests of phonological short-term memory, in which people with WS are impaired, involve stimuli of 2, 3, 4 and 5 syllables. Additional supports for language acquisition must be available because the vocabulary ability of WS subjects can reach a level much higher than that of a normal four-year-old. It may be that a system of rote learning is used to build up the lexicon. Indeed, it has been shown that WS subjects do not use previous word knowledge to support new word repetition (Grant et al., 1997). This is further evidence in support of some form of semantic deficit in WS.

4.3.2 Mental age and vocabulary acquisition

Despite having a relatively large lexicon in later life, children with WS acquire language later than their normal counterparts. In addition, the sequence of language acquisition is different from that in normal development, as demonstrated in a study by Thal, Bates and Bellugi (1989). They studied the relationship between early language ability and symbolic representations in two young girls with WS. The two girls, Becky (5 years 6 months) and Rachel (23 months), were not yet combining words and, alongside this, they did not follow the normal pattern in the relationship between language and gesture. In normal development, similar abilities appear in both language and gesture at the same time. Thus when typically developing children begin combining gestures to form sequences at 20-24 months, they simultaneously begin combining words (Bates, Benigni, Bretherton, Camaion & Volterra, 1977). However, for the two children with WS the pattern of single words versus single gestures and combining of words and combining of gestures turned out to be different from the normal pattern. Although Becky and Rachel had poor vocabularies, in several gesture tasks they performed above their language ability, if the appropriate support was given. Becky was able to perform single gestures as easily as normal children who were combining words. However, in combining gestures (for example, putting a teddy to bed in a sequence of stages), both the girls performed below their language level. It appears that they were using different strategies to perform the gesture tasks, and that this may have been influencing acquisition. They seem to rely heavily on contextual cues in the tasks. For example, they succeed on tasks when real objects are used (e.g. a toy telephone) but not when placeholders are used (e.g. a banana that could be used in place of a real telephone). Their problem with sequences of gestures may be due to taking the sequence as a whole indivisible unit, rather than dividing it up into parts and repeating them one by one. This same approach could be reflected in their language acquisition. It has been argued that the relative strength of language in WS could be due to a rote learning strategy (learning words as wholes) rather than a more rule governed, analytic approach as is seen in most young children as they develop (Karmiloff-Smith, Grant, Berthoud, Davies, Howlin & Udwin, 1997).

4.3.3 Language processing: brain imaging techniques

It would also be crucial to examine the neural correlates of lexical behaviour in infants with WS as is being done with normal infants. However, at present no studies have been undertaken with infants. Adults with WS have been shown to exhibit abnormal event related potentials (ERPs) to sound stimuli, perhaps due to the preponderance of hyperacusis in this population (Neville, Mills & Bellugi, 1994). ERPs were also recorded during semantic priming experiments. It seems that adults with WS do not process sentence meaning in the normal way, which may suggest a different developmental pathway. The latter data are particularly relevant to infancy work. Different linguistic processing in later life may suggest different beginnings in infancy. When WS adults and children (from 8 years old) were presented with words aurally, they showed a far greater P200 response than normal age-matched controls. The participants with WS and controls were also presented with sentences which had either expected endings (e.g. I take my coffee with cream and *sugar*) or unexpected endings (e.g. I take my coffee with cream and *radiator*). The WS group show an abnormally large positivity to the expected ending than the controls (at any age) and this suggests there may be a greater priming effect in the auditory modality than in age matched normals (Neville, Holcomb & Mills, 1989; Neville et al., 1994). This large priming effect suggests that there may be stronger interconnections between lexical items in WS subjects and could point to a differently structured lexicon in adults. This, in turn, could mean that the lexicon developed in a different manner. These facts hold for aural presentation, By contrast, for tasks presented visually to those who could read, the WS group showed a normal ERP response. In the written semantic priming task, responses were the same as those for controls.

There is also tentative evidence to suggest that the organisation of language within the hemispheres may differ somewhat in WS. Unlike normal controls, they do not show increased priming effects to visual or auditory stimuli in the right hemisphere (Neville, Mills & Bellugi, 1994). It may be that the late acquisition of spoken language and reading has altered this pattern, adding weight to the argument that the timing of developmental events has a profound effect on subsequent brain organisation.

The following section outlining within-language differences in infants and young children with WS will be necessarily brief. Very little research has yet been carried out with younger WS individuals.

4.3.4 Language comprehension and production: a dissociation of abilities?

There is a small amount of research that characterises the pattern of production and comprehension abilities seen in Williams Syndrome. Using data from the MacArthur Communicative Development Inventory (CDI), a language questionnaire that is completed by parents, Singer Harris, Bellugi, Bates, Jones and Rossen (1997) compared the comprehension and production of 39 infants and children with DS and 54 with WS.

This will be discussed more fully in the CDI study, but at this stage it is important to note, that compared to the norms for this scale, both production and comprehension are delayed in infants with WS. This is also evident in languages other than English. In a study of Italian children from 4 to 15 years of age, lexical comprehension appears to be better than production. The participants in Volterra, Capirci, Pezzini, Sabbadini and Vicari's study (1996) showed poor production on the Boston Naming Test, but understood more words than would be expected given their MA. This is supported for the English language by data from two English speaking girls aged 23 months and 5;6 years (Thal, Bates and Bellugi, 1989).

In sum, more data need to be collected from atypical infants who are in the very earliest stages of language acquisition. The data from somewhat older children with WS suggest that vocabulary is delayed and that, as in typical development, comprehension ability outstrips production.

4.3.5 Lexical and syntactic comprehension: a dissociation of abilities?

In the section on typical development, the asymmetry between lexical comprehension and the comprehension of syntax was described. Is this asymmetry also present in the language development of infants and toddlers with WS? Singer Harris, Bellugi, Bates, Jones and Rossen (1997), began to address the question of syntactic production, but not comprehension, using data from the MacArthur Communicative Development Inventory (CDI). The CDI comes in two forms and it is the second (Words and Sentences scale) that is of interest here because this begins to assess the emergence of grammar. Grammar was assessed in two ways. One of these was length, in which the mean length of the 3 longest utterances was taken. The other was complexity, in which parents were presented with pairs of sentences and were required to indicate which sentence in each pair was closest to those their child could produce. For infants with a mean CA of 47 months, Singer Harris et al. (1997) found that those with WS were producing more advanced grammatical structures, in terms of both length and complexity, than the DS group. This was despite the fact that single word production was equivalent in both groups. Despite delay, once the WS group began using grammar, they appeared to be following the normal pattern of development. These infants produced utterances of equivalent length and complexity to typically developing infants who were at the same level of lexical production. This finding needs to be considered in the light of data gathered from older individuals. Older children and adults with WS display several problems with complex grammar (Karmiloff-Smith et al., 1997). The discrepancy may be due to the nature of early grammatical development. The WS adult population has problems with structures that may not yet be present in the young children's grammatical constructions.

There are also data on language comprehension from Italian children with WS. Four to 15 year olds do not differ from MA-matched controls on the Peabody Picture Vocabulary test that assesses lexical comprehension (Volterra et al., 1996). However, these same children perform poorly compared with controls on the Test for Reception of Grammar, a measure of grammatical understanding. WS children make the same types of errors as the MA controls, suggesting that development is delayed not

different. However, a case study of a girl followed from the age of 2;6 to 4;10 provides data on productive syntax and morphology which suggests deviance as well as delay (Capirci et al., 1996).

4.3.6 Summary

The studies described above concerning WS language development only begin to address questions that have been more thoroughly considered in the DS literature. However, both the behavioural evidence from younger children with WS, and brain imaging data from older children and adults collected thus far suggests that the lexicon develops differently in WS and that there are also problems with syntactic processing. These impairments are evident despite relatively good vocabulary levels when compared with mental age. Further studies examining lexical acquisition in infants with WS should indicate the different pathway that lexical development takes in this population, and may explain how the relative strength is supported.

4.4 Literature summary

The review of the literature presented above has pointed to clear differences in the way WS and DS language develops, despite language delay in both groups. The evidence points to a delay in the development of both vocabulary and syntax in the DS group. However, it is also apparent that this group follows the typical developmental pathway and achieves important milestones which link language and cognition in the same sequences as their typically developing counterparts. In contrast, the WS group does not follow the same developmental trajectory. This is evident in both the sequence in which they reach different milestones and the fact that they do not make use of some of the constraints which guide the language acquisition of the other groups.

The following empirical studies will examine language performance across both the WS and DS groups, to see whether adult performance is reflected in the starting states of these syndromes, and how the performance across the groups differs.

Part Two: Empirical Studies

4.5 Experiment One: Vocabulary Comprehension and Production - Data from the MacArthur Communicative Development Inventory

4.5.1 Introduction

Singer Harris et al. (1997) used the MacArthur CDI questionnaire to assess the vocabularies of infants and children with DS and WS. Data from the words and gestures scale were analysed for 34 infants with WS and 32 infants with DS, mean chronological ages 34 and 32 months respectively. Both the DS and WS infants' production and comprehension was delayed by about 15 months, but the relationship between these abilities was similar to that found in typically developing infants. They demonstrated the expected discrepancy between comprehension and production ability, as seen in typical development (Bates et al., 1988; Fenson et al., 1994). In addition to this analysis, Singer Harris and colleagues divided the sample into two groups: those children with comprehension vocabularies above 50 words and those below 50. There was no difference in the abilities of the DS and WS groups for participants with vocabularies above 50 words. However, an interesting difference was found for the participants with smaller vocabularies. The DS group understood more words than the WS group, a surprising finding given the WS language advantage in later life. The use of gestures, an early indicator of language development, was also assessed. The DS group produced an unusually high number of gestures in relation to vocabulary, something that has been reported widely (Caselli et al., 1998; Miller, 1987). This may well be linked to the wide use of gestural communication in children with DS and may also reflect a reliance on gesture as a compensatory strategy for the lack of spoken language.

The present study aims to characterise early comprehension and productive vocabulary, using the same measure as Singer Harris et al. (1997) and to subsequently

compare experimental data in these same infants. Given the adult literature, it is predicted that the DS group will perform poorly in comparison with the WS group, especially on production items. Both atypically developing groups are expected to be delayed relative to CA and MA-matched groups. It will be interesting to see whether the surprising comprehension advantage present in Singer Harris et al.'s DS group at the very beginning of vocabulary is replicated in the data from my sample. Singer Harris and her colleagues suggest that their finding may be due to that fact the some of their WS participants did not exhibit "classic" WS symptoms because many of the data points came from older children who were still at the very beginning of language acquisition. It is unclear what this "classic" form of WS might be. However, a further investigation may clarify this uncertainty about WS and DS performance very early in language development.

Although the present study used the same populations and measure as in the Singer Harris et al. study, it differs in several important ways. First, it was conducted using younger participants who spanned a much narrower age range, from 24 to 36 months. By contrast, in the previous study the infants ranged from about 10-55 months old. My study can therefore provide information about a much better defined point in development. In addition, my infants came from a pre-existing sample in which infants with WS and DS had been matched on sex, overall mental age and as far as possible socio-economic status. Those in the previous study only provided CDI data so it was rather difficult to make more general comparisons across the groups.

4.5.2 Method

Participants

All parents whose infants took part in the research program were asked to complete the MacArthur CDI. Despite the fact that parents were sent questionnaires to complete before their visit, not all were returned. Data were obtained from 49 infants, yielding 56 cases. Eleven infants with Williams Syndrome contributed data. Five contributed data twice (at an interval of 6 months) and six contributed once, yielding 16 cases. Eleven infants with Down Syndrome contributed data. Two contributed data twice (at

an interval of 6 months) and nine contributed once, yielding 13 cases. In addition, 16 MA-matched normal infants and 11 CA-matched normal infants were tested. See Table 4.1 for a summary of the chronological and mental ages of the participants.

Table 4.1 Mean chronological and mental ages for each group

	Mean CA (months)	SD	Range (months)	Mean mental age (months)	SD	Range (months)
WS	30	4.90	24-36	16.06	2.82	11-21
DS	28	4.34	24-36	14.6	2.06	12-19
MA-matched	15.13	2.42	12-20	15.19	2.74	11-21
CA-matched	31.09	5.24	24-36	31.64	5.20	26-40

Materials

The MacArthur Communicative Development Inventory (CDI): Words and Gestures form was used. This parental questionnaire allows for assessment of both vocabulary comprehension and production as well as the understanding of simple phrases and use of gestures. For the present study, only the responses to the 396 vocabulary items were considered. The infant version of this scale was designed for use with typically developing infants up to 16 months. It was decided to use this version, not the chronological age appropriate scale, because the infants in my sample were not yet combining words and the toddler scale has a large section dedicated to early grammar.

The high validity of parental report using the CDI has been reported on a number of occasions, both for typically (Fenson, Dale, Reznick, Bates, Thal & Pethick, 1994) and atypically developing infants (Miller, 1992b; Miller, Sedey & Miolo, 1995). The measure correlates highly with observational data and with later language ability. This makes the CDI a convenient measure to use, enabling the collection of a large amount of data in a relatively short time.

Procedure

Parents were asked to indicate whether their child: i) understood or ii) understood and said a particular item on the questionnaire. Vocabulary items were divided into 19 categories. These were: sound effects and animal sounds, animal names, vehicles, toys, food and drink, clothing, body parts, furniture and rooms, small household items, outside things and places to go, people, games and routines, verbs, adverbs of time, adjectives, pronouns, interrogatives, prepositions and locations, and quantifiers. Items were chosen because they are some of the first to appear in typically developing children's vocabularies. Parents were told to consider the British English equivalent for Americanisms, such as diaper, whenever they occurred.

4.5.3 Results

Data analysis

The percentage of items understood and the percentage of items produced was calculated for each infant:

Mean number of words comprehended (produced)

Total number of words (396)

Overall results

A repeated measures Anova was conducted, with group (WS, DS, MA-matched, CA-matched) as the between-subjects factors and type of response (production or comprehension) as the within-subjects factor. (See the footnote on page 43 for a discussion of the use of multiple data points from individual participants.) As would be expected given the literature on typical development, all groups were able to understand more words than they could produce, $F(1,52) = 109.96$, $p < .001$. Readers interested in data from individual participants can find this tabulated in Appendix D. These tables show that production scores are greater in the WS group than in the DS group. This is true even if the older (36 month old) toddlers with WS are removed.

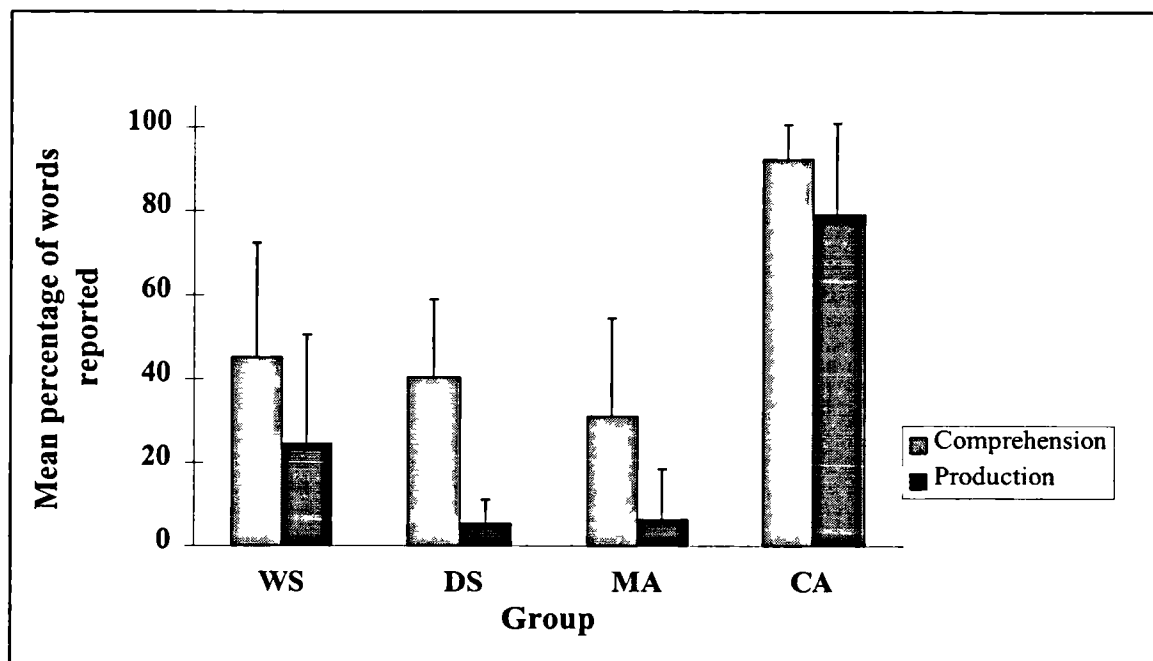
Differences between groups

There was an effect of group, $F(3,52)=34.29$, $p<.001$. The CA-matched group produced and understood a larger number of the items on the CDI, as demonstrated in Figure 4.1 for the mean percentage scores for each group. This would be expected as the version of the questionnaire given in this study was standardised for infants up to 16 months, half the mean age of the CA group. The MA-matched group performed at the level of their chronological age, as defined in the CDI manual. The atypically developing groups understood more items than their MA-matched counterparts, though this difference was not significant (Tukey HSD, $p>.05$). This trend could be due to the fact these groups are older and therefore have had more opportunity to increase their vocabulary due to their greater experience of language.

If the WS and DS groups are considered in isolation, using a one way Anova, there is a significant difference between the comprehension/production discrepancy, $F(1,27)=6.13$, $p<.05$. The difference in the WS group is significantly smaller than that seen in the DS group, as discussed below.

In addition to effects of group and type of response, there was a significant interaction between the two, $F(3,52)=3.77$, $p<.05$. This meant that the pattern of abilities varied with group, as presented in Figure 4.1, which shows differences between the comprehension and production scores of each group. The differences across groups seem to lie in performance on production items. Post hoc tests (Tukey HSD) reveal that all groups except the DS and MA-matched groups differ on this measure $p<.05$ and $p>.05$ respectively. The only difference seen in the production data was that between the DS and CA-matched group (Tukey HSD $p<.05$), revealing that DS production is well below that expected for their chronological age.

Figure 4.1 Performance on the CDI: comprehension and production



Performance of the Williams Syndrome group

When the data are presented graphically (see Figure 4.1), the pattern in the Williams Syndrome data appears similar to that in the data from the CA-matched. However, a post hoc analysis of the differences between scores between each group revealed that the WS pattern was not different from the other three (the CA-matched group pattern, Tukey HSD, $p < .05$ or the MA-matched pattern or the DS group pattern). Infants with WS understood significantly more words than they produced, post hoc t test ($df = 15$) $= 5.58$, $p < .001$, as would be expected in normal development. However, the number of words understood and produced in the WS was much lower than in CA-matched infants (Tukey HSD, $p < .05$), but higher than that for MA-matched infants. Therefore, the pattern seen in the WS group is neither like that of a child of the same age nor a child of the same mental ability. This would suggest that language in individuals with WS follows a different developmental trajectory.

Performance of the Down's Syndrome group

Like the WS infants and the 2 control groups, the Down's Syndrome group produced less words than they understood (post hoc t test $t(df\ 13) = 7.58, p < .001$). But the discrepancy between comprehension and production was larger than that in the WS group and may be at the root of the language problems that people with Down's Syndrome experience throughout their lives. These data certainly support the claim that there is a distinct production/comprehension discrepancy in DS (e.g. Miller 1987, 1989). The DS group does not differ from the MA-matched group in terms of productive or receptive vocabulary. This means that vocabulary is at the same level as general cognitive ability in the DS group, suggesting language delay rather than deviance.

4.5.4 Discussion

The data from this parental assessment of language ability reveal some interesting differences between DS and WS. It appears that the DS group are following a similar trajectory to that seen in normal development, as they are performing in a manner very much like that of children with the same mental age. In contrast, despite significant language delay, the WS infants do not share the same profile as MA-matched infants. Neither is the pattern similar to that of infants at the same chronological age. This fits with existing data from other aspects of language development (Mervis et al., 1997). For example, as mentioned earlier, data on the emergence of referential pointing behaviour indicates that in DS pointing comes before labelling as in typical development (Harris, Barlow-Brown & Chasin, 1995) but that in WS pointing emerges well after the infant produces his first words. The unusual profile highlighted by the present study may be a glimpse of the beginning of the atypical developmental trajectory that WS language development will follow and merits further systematic research.

Several investigators have suggested possible reasons for the production difficulties found in individuals with DS. We know that children and adults with DS have poor verbal memory ability (Buckley, 1997; Marcell & Armstrong, 1982) and that in typical development verbal memory is an extremely important tool for vocabulary

acquisition (Gathercole & Baddeley, 1990 a,b). However, it would appear that speech motor problems are a more likely cause of the production deficit in the DS group, given their relatively spared comprehension ability. Verbal memory problems would affect both production and comprehension, whereas speech-motor problems would have a greater affect on production.

The present investigation did not find that the DS group showed a comprehension advantage over the WS group in the early stages of language development as suggested by the Singer et al. data. Neither did the data indicate that the WS group exhibited significantly better language comprehension than the DS group, as the adult literature would suggest. Both groups' comprehension was similarly impaired. In addition, the DS group was particularly disadvantaged on the production measure. This would suggest a crucial point: that differences seen in these groups' language abilities in adulthood arise as a result of development. The DS production problems displayed in infancy may increase as development progresses. This group might find it hard to progress beyond simple language production and encounter difficulties with multi-word speech. In order to discover the causes of the comprehension impairment in both groups and the particular problems with production in DS, it is necessary to carry out further studies.

4.5.5 Conclusions

This study has shown that there are differences in the language profile of the DS and WS groups even at this very early stage of language development. The DS infant profile is delayed, whereas the profile seen in the WS group does not fit with profiles seen in either their mental age or chronological age-matched counterparts. The data emphasise the importance of characterising the language profile early in development. Although language in WS appears relatively spared in adulthood, the data discussed here suggest that their language is not a product of the normal processes of language development.

4.6 Experiment Two: Do infants understand single nouns?

4.6.1 Introduction

Although validation of parental reports has been carried out for normally developing children (Fenson et al., 1994), it remains possible that parents of atypically developing children may under or over-estimate their children's linguistic competence. Experiment two: - set out to assess the single word comprehension of the infants using a laboratory methodology rather than parental report. If the results from a controlled experiment mirrored those from the CDI, then this would add further weight to the reliability of parental assessment of language ability in atypical groups.

A preferential looking paradigm was chosen to address the question of single word comprehension. This method has been used successfully with typically developing children (Golinkoff, Hirsh-Pasek, Cauley & Gordon, 1987) and is particularly suited to participants with Down's Syndrome and Williams Syndrome because of the low task demands. Comprehension is assessed by measuring looking time to visual stimuli. The infant is presented with two images while simultaneously hearing a linguistic stimulus that refers to one of the images. Research into intermodal perception has shown that infants will look longer at the stimulus which matches the auditory event (Spelke, 1976, 1979). For example, the infant tends to look longer at a jumping stimulus when presented with "landing" sounds. The task requires no speech production and very little motor activity (only eye movements). The following section will describe what is known about single word comprehension in both typical and atypical development using various experimental paradigms.

Single word comprehension in typical development

Before Golinkoff and her colleagues (1987) used the preferential looking paradigm to assess single word comprehension in infancy, many of the comprehension tasks used were much more demanding. Children under two years old were asked to point to objects or pictures, or to manipulate toys. These differing task demands meant that the results from such studies were often discrepant. Some researchers reported that lexical

comprehension appears before production (Nelson, 1973), whereas others found comprehension to lag behind production (Chapman, 1977). This is an essential issue for atypical development. It is important to discover whether atypical language develops normally, but is delayed, or whether it follows a deviant pathway. If one can characterise properly the order of acquisition in normal development, this will then provide a benchmark for the atypical case. Discrepant results could arise for a number of reasons. It could be that former comprehension studies assessed not only linguistic ability but also task understanding and co-operation (Bloom and Lahey, 1978). With these problems in mind, Thomas, Campos, Shucard, Ransay and Shucard (1981) tested comprehension using a signal detection procedure. Infants as young as 13 months looked longer at objects that were labelled with the object name than those labelled with a nonce word or a word not referring to the object. Cardoso-Martins, Mervis, and Mervis (1985) and Woodward, Markman and Fitzsimmons (1994) have also shown referential comprehension in 13 month olds. It seems that this ability emerges sometime in the second year, given that two studies with 11 month olds have been unsuccessful (Thomas et al., 1989; Behrend, 1987).

In a preferential looking study, similar to the one reported below, Golinkoff et al. (1987) showed that typically developing 16 month old infants are able to link a word to its referent, demonstrating this by looking longer at the stimulus which matches the linguistic input. Despite these results, they add a caveat. The infants performed better on the first three trials than on the last three trials. It is suggested that the infants fell into a distinctive looking pattern, in which they favoured the image on the side that matched the linguistic input on the preceding trial. This meant that even when they shifted their gaze to the correct side the total looking times to each image were quite even and therefore for those trials no statistical differences could be found (Hirsh-Pasek, Golinkoff, Beaubien, Fletcher & Cauley, 1985). However, infants never looked solely at one side. It is also important to note that results from these studies suggest comprehension levels are higher than those reported by parents (Hirsh-Pasek et al., 1985).

The results described above suggest that, interestingly, the pre-verbal infant, or infant just starting to produce language, is able to understand single nouns. The results have

been largely due to the preferential looking paradigm employed by Golinkoff and her colleagues. This is an exciting technique and is a useful tool in answering many questions about early language development. A preferential looking paradigm is used in the study reported below. Following on from previous results using the technique, it is hypothesised that both the CA and MA-matched groups in the study would look longer at the matching than the non-matching visual stimulus, indicating single word comprehension.

Single word comprehension in Williams Syndrome

Previous studies of comprehension in infants with WS have been limited to the use of the MacArthur CDI or observation methods to investigate both understanding and production, as discussed earlier. The following study goes beyond this and is the first experimental assessment of lexical comprehension in infants with WS. Some previous anecdotal reports suggest that individuals with Williams Syndrome may produce more language than they understand (see Singer-Harris et al., 1997, for a discussion). This means that they could use words but be unaware of their meaning. This might suggest that comprehension does not precede production in WS infants. The following study can begin address this suggestion by examining whether infants who are producing little or no language are able to understand single words. This is possible because comprehension skills of participants who are producing little or no language can be examined using preferential looking methods. If the group do comprehend single words, this would suggest that comprehension is a precursor to production as in normal development. Given the superior language abilities of individuals with WS in adulthood, it is predicted that comprehension in this group may be better than that found in the Down's Syndrome group even at this early stage in development. However, if the infant phenotype cannot be inferred directly from the adult pattern, then this may not hold.

Single word comprehension in Down's Syndrome

Mervis and colleagues (Cardoso-Martins, Mervis, & Mervis, 1985) found that infants with DS begin to understand their first words at about the same mental age as their

typically developing peers, around 14 months. This was done by presenting the infants with 4 toys and asking them to point out the one asked for by the experimenter. Between the mental ages of 13-21 months, infants with DS have similar sized comprehension vocabularies to their MA-matched counterparts, suggesting that the speed of vocabulary growth is normal even if onset is seriously delayed. Several studies have shown that language comprehension in DS is at a similar level to their general cognitive abilities (Miller, 1996; Stella, Lami, Caselli, Casadio & Pizzoli, 1993). However, no preferential looking studies investigating word understanding have yet been conducted. The present study will be the first to employ the preferential looking technique and should be able to tap the very beginnings of language comprehension in this group, perhaps missed by other techniques. The infants are not required to make any complex motor responses or vocalisations to the stimuli in this task whereas the high demands in other studies may have affected their performance. In spite of the lower task demands in the following study, it is nevertheless predicted that infants with DS may exhibit lower levels of comprehension than typically developing infants or those with WS, given the poor language skills of many adults with DS.

4.6.2 Method

Participants

Data were collected from 71 infants: 15 with WS, 22 with DS, 17 mental age-matched controls and 17 chronological age-matched controls. (Of the 71 infants, 63 also participated in Experiment One in Chapter 6.) See Table 4.2 for details.

Design

A preferential looking paradigm based on Golinkoff, Hirsh-Pasek, Cauley and Gordon (1987) was used. Each infant was presented with 16 trials made up of a pair of images, each seen twice.

Table 4.2 Mean chronological and mental ages for each group

	Mean CA (months)	SD	Range (months)	Mean mental age (months)	SD	Range (months)
WS	30.4	4.79	24-36	16.5	2.39	12-20
DS	29.7	5.06	24-36	15.6	2.36	12-20
MA-matched	15.4	2.33	12-20	15.1	2.48	12-21
CA-matched	30	5.13	24-36	30.6	5.14	25-40

Materials

Infants were tested using a replica of the basic Fagan apparatus (Fagan, 1970), as shown in Figure 4.2. This apparatus is a portable visual-preference viewing box, which allows the presentation of two visual stimuli simultaneously. It has a hinged display panel (85 cm long and 29 cm high), with two slots to hold the stimulus cards, as in Figure 4.3. In this study, the stage was illuminated using a fluorescent lamp, which was out of the infant's view. The position of the lighting enabled the experimenter to see the infant's pupils clearly. The centre-to-centre distance between the slots was 30.5 cm, and on all trials, the display stage was situated approximately 30.5 cm above the infant's head. In the centre of the stage was a peephole 0.625cm in diameter, through which an observer, blind to the position of the stimuli, could see the visual fixations of the infant. Each infant was tested in a special infant seat in order to keep them still and in the correct position.

The stimuli were coloured photographs mounted onto white cards 17.7 x 17.7 cm. Photographs were used because parents reported that infants with Down's Syndrome react more readily to a photograph than to a drawing. Images of 16 familiar objects were chosen and their size, brightness and complexity was controlled as far as possible. The following objects were used: cup, clock, table, house, spoon, phone, teddy, doll, ball, socks, shoes, hat, cat, dog, duck and fish. Stimuli were presented in pairs that were randomly generated. Every pair was seen twice, with the position of the cards changed on each occasion.

Figure 4.2 The Fagan Box

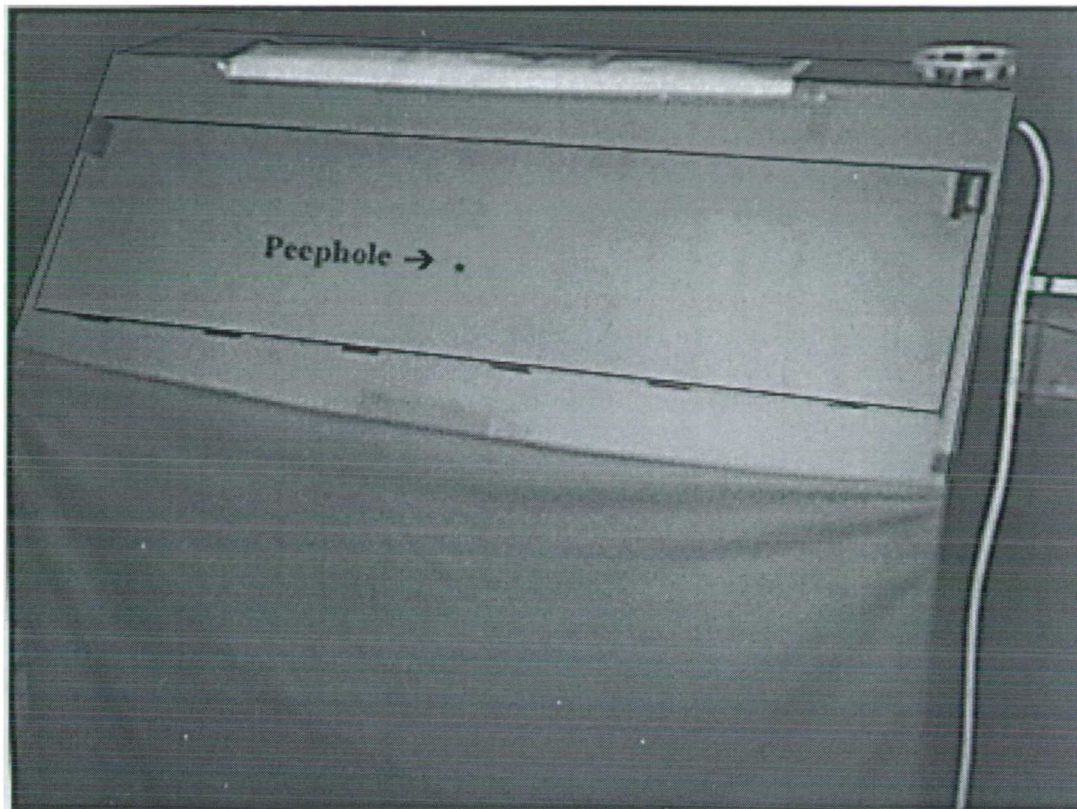
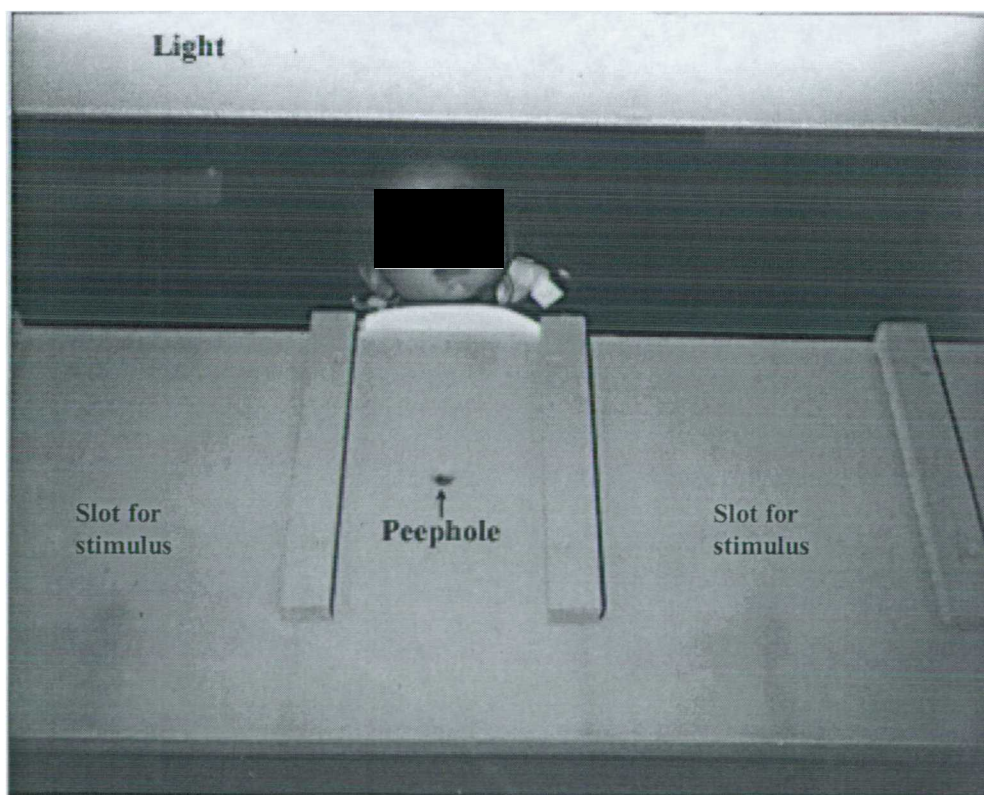


Figure 4.3 Fagan Box from above



Procedure

The child was settled into the infant seat and then the testing apparatus was wheeled into position, with the peephole in the display stage centred directly over the infant. During this time, the experimenter spoke to the infant, who could no longer see the parent. The stimuli were then placed into the two slots simultaneously by the experimenter. Once the infant's attention was obtained by talking or by shaking a rattle, the presentation began. Just before presenting the display to the infant, i.e., just as the flap was closing, the experimenter began the linguistic input. In a loud, clear voice, using the highly pitched intonation of "motherese" she said, "Look, look at the X" (the named stimulus) and repeated this 3 times while the stimuli were on display. Each infant was shown 16 trials. Each trial consisted of the presentation of two pictures. Each pair was presented twice, once with the named item on the left and again later in the sequence with the named item on the right, or vice versa. The side on which the named object appeared was randomised and the observer, who recorded the looking time, was blind to the position of that card. She held a stopwatch in each hand and timed the infant's looking to the left versus the right stimulus item by observing the corneal reflection of each stimulus in the infant's pupil. Reliability using this procedure has been shown to be high (Haaf, Brewster, de Saint Victor & Smith, 1989; O'Neill, Jacobson & Jacobson, 1994).

A timer, which was set to 5 seconds, signalled when a trial was to end. Between each familiarisation trial, the experimenter pulled back the flap from the infant's view, recorded the times called out by observer, and changed the stimulus cards. Once the infant's gaze was centred and her attention obtained, the flap was closed exposing the next stimuli to the infant.

4.6.3 Results

Cumulative looking time to each picture was recorded by the experimenter. Only data from pairs in which the child was familiar with both objects, as assessed by parents before the test, were included in the analysis. This was done following a problem with the Golinkoff et al. (1987) study. Golinkoff and her colleagues point out that if an

infant is presented with two images, e.g. sock and hat and is familiar with only one of them (sock), he might look at the item corresponding to the verbal input (hat), simply because he knows a sock is not a hat. The number of items included in the analysis varied from two pairs ($n=2$ MA controls) to all 16 pairs, depending on the parental report.

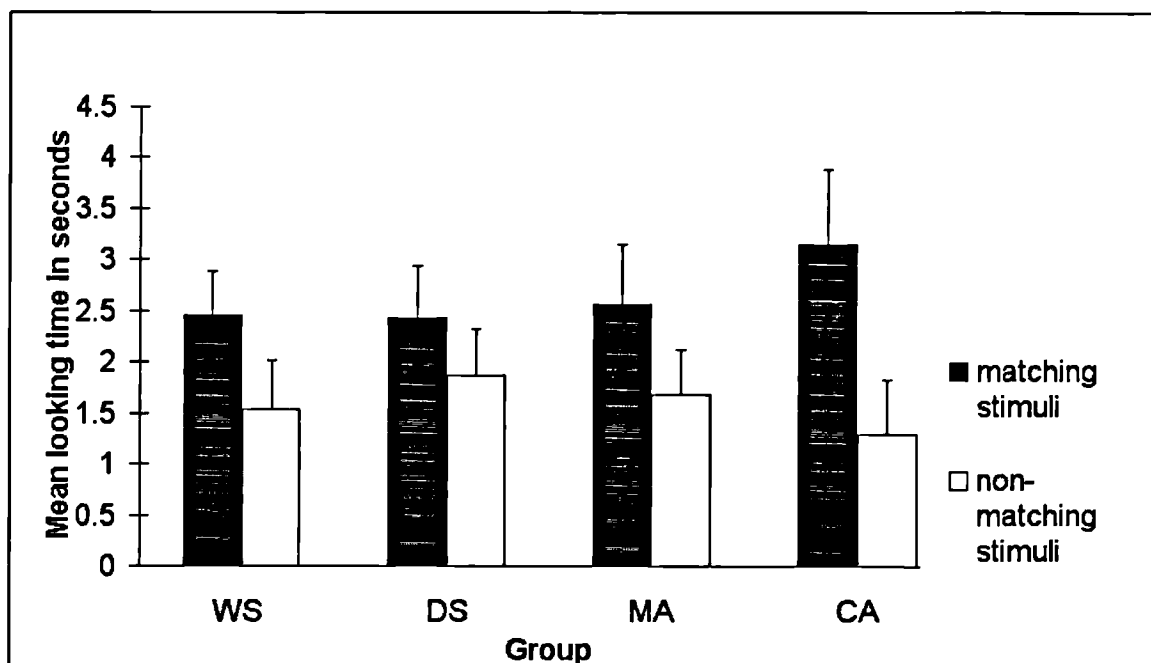
If an infant had formed a link between a spoken word and its referent, one would expect that she would look longer at the picture that matched the label which she heard than at the non-matching picture. Analyses were carried out to ascertain whether infants in each group exhibited this understanding.

Not all infants were included in the analyses. Outliers were found using a box and whisker plot. The main body of the plot, represented values between the 25th and 75th percentile. Participants whose looking times were more than 1.5 box lengths above or below these values were excluded from the analysis, according to the instructions provided in the SPSS statistical package (Kinnear & Gray, 1999). This made very little difference to the numbers in each group. One infant was removed from both the WS and MA-control groups and two were removed from the DS group. There were no outliers in the CA-control group. These removals made no difference to the DS or WS results. The infant MA-control who was removed was not attending to the task. If this data was included the MA results would still show a trend in the appropriate direction.

A repeated measures Anova with group as the between-subjects variable and match or non-match as the within-subjects variables, was carried out to identify differences in mean looking times across subjects. The mean looking times to matching and non-matching stimuli by group are shown in Figure 4.4. There was a significant difference in looking time to matching and non-matching stimuli, $F(1,64) = 87.79$, $p < .001$ with longer looks to the matching stimulus. No effect of group emerged, $F(3,64) = 1.90$, n.s. However, there was a significant interaction between group and match, $F(3,64) = 6.64$, $p < .001$. Post hoc t-tests examining the differences within each group between looking times to the matching and non-matching stimuli revealed significant

differences in all groups, but to a lesser extent in DS (WS $t(df\ 13) = 4.65, p < .001$; DS $t(df\ 20) = 3.21, p < .004$; MA-control $t(df\ 15) = 4.06, p < .001$; CA-control $t(df\ 16) = 6.41, p < .001$). A one way Anova was carried out to examine whether the magnitude of these differences varied between groups, with difference (matching stimuli minus non-matching stimuli) as the dependent variable. There was a significant difference $F(3,67) = 5.58, p < .001$. Post hoc tests were then carried out to compare the magnitude of the difference between looking times to matching and non-matching stimuli for each of the groups. The CA group was significantly different from all other infant groups (Tukey HSD, $p < .05$). There was no difference between the WS and DS infants.

Figure 4.4 Mean looking times to matching and non-matching stimuli in seconds



4.6.4 Discussion

The results show that even in the very earliest stages of language acquisition it is possible to assess the level of comprehension of atypical infants using a preferential looking paradigm. In my study, both typically and atypically developing infants were

able to make a link between the spoken word and its referent, looking longer at matching than non-matching stimuli.

This difference in looking time was the smallest for the Down's Syndrome group but nonetheless significant. It may be that the group had to look more carefully at both stimuli before settling on the matching item. If this is so, then the difference in looking time to the two stimuli would be less marked. This said, the findings show that all the infants could do the task.

4.6.5 Conclusions

Despite marked differences in later language ability, both the DS and WS are able to match a word to its referent at this early stage in development, i.e. 24-36 months, well before they are producing language. Again, this highlights the importance of studying language ability in infancy, particularly in clinical groups. The pattern of language abilities seen in the DS and WS groups in this infant study is not the same as that seen in adulthood. This would have been missed if infant performance had been inferred from adult data, because the adult data suggest that WS vocabulary comprehension should be higher than that present in their counterparts with DS. The adult data also give little indication of the degree of delay in both groups.

The fact that the Williams Syndrome and Down's Syndrome groups exhibited this level of comprehension suggests that these individuals are capable of understanding language before they are able to produce many, if any, words at all. In this respect, then, both groups appear to be acquiring language like their typically developing counterparts. However, the more even pattern of looks to matching and non-matching present in the DS data might be a first sign of their future language difficulties. This must however be investigated further. A task with more items might clarify the difference in the WS and DS looking patterns, by providing more scope for differing degrees of ability.

4.7 Experiment Three: Do infants understand the importance of word order?

The experiments reported thus far have shown that infants with DS and WS are able to understand common nouns in isolation. Although the DS group has a marked delay in productive vocabulary, both groups display a level of lexical understanding appropriate for their mental age. A further preliminary study was carried out to build upon the evidence of single word understanding, with an investigation of the early stages of grammatical comprehension. Grammar is a particularly interesting aspect of atypical language development. In adulthood, individuals with DS have particular difficulty with both productive and receptive grammar. These difficulties are far more marked than those seen in vocabulary. Despite relatively good language compared with DS adults, adults with WS also have grammatical difficulties. Several studies point to certain impairments in morpho-syntax (Karmiloff-Smith, Tyler, Voice & Sims, 1998; Karmiloff-Smith, Grant, Berthoud, Davies, Howlin & Udwin, 1997). Given these outcomes, it is important to characterise grammatical understanding early in development.

Word order was chosen as the candidate structure for the study because it is one of the first grammatical forms to appear in English speaking children's speech (Bloom, 1970; Brown, 1973). It is therefore likely to be something to which infants hearing English pay special attention. Several studies have examined word order in typical development. These provide a base from which to investigate this aspect of grammar in WS and DS infants.

This was a pilot study because only a very small group of infants took part. It used the preferential looking technique to examine sensitivity to word order (Hirsh-Pasek & Golinkoff, 1996). The sample was small because infants were often fussy when this study was conducted, as it was the last item tested in the Fagan box. Testing was only carried out if the infant was still happy to remain in the apparatus.

In the study infants were familiarised with two characters (Cat and Duck) and then presented with two stimuli. In one, Cat was acting upon Duck, e.g. Cat pushing Duck,

and in the other, Duck was acting upon cat. These stimuli were accompanied by a simple sentence, e.g., “Look, Cat is pushing Duck”. The infant cannot rely simply on the character labels to pick which stimulus corresponds to the verbal input because both characters appear in both stimuli. Instead, infants must attend to word order to make their decision. Four verbs were tested: feed, push, wash and hug, and infants were presented with stimuli for each verb four times, so that mean looking time to each stimulus could be calculated.

Because of the small sample sizes, it is not possible to evaluate word order comprehension in DS or WS. It is necessary to collect complete data from many more infants before firm conclusions can be made. Despite this, the study did suggest that the oldest CA-matched infants do make use of word order information when focusing on the appropriate picture to match verbal input. Further studies, using dynamic video stimuli to increase verb salience and to capture the infants’ attention, are planned in order to address this question, with a separate familiarisation phase, prior to the preferential looking phase.

4.8 Further avenues for research

These were the first experimental studies of their kind to be conducted with atypically developing infants. Because of this, certain problems arose with the methodology that could be readily solved in further experiments. The most important of these were the design and type of stimuli used in the word order study. As discussed above, task demands should be lowered by introducing a familiarisation phase to increase the probability of character identification by atypically developing infants. Without the ability to identify the characters readily, it is of course not possible to assess the infant’s understanding of word order. It is also important to present moving stimuli to the infants so that the agent and patient of the verb are very clear. Unfortunately, it was not possible to present moving stimuli at the time that this study was run. The study will be conducted again in the future using the new video-based preferential looking set-up in our laboratory. Further research could investigate other constructions, in particular transitive and intransitive sentence frames which have

been successfully studied in typically developing 27 month olds (Hirsh-Pasek et al. 1988) and have been shown to be impaired using more complex stimuli with adults with WS (Karmiloff-Smith et al., 1998).

The use of the CDI was successful, but in future studies some amendments could be made. Children with DS are often exposed to Makaton signs (e.g., Clibbens, 1993) and the inclusion of their knowledge of signs in the analysis of these data might be interesting. It is likely that production levels would increase if signs were included, and therefore underlying language ability and not articulatory motor deficits might be assessed. However, if making a comparison with typical development, then the normal means of communication, i.e. speech, are of greater interest and should be measured so that two different analyses can be carried out. It has also been noted (Mervis, personal communication) that if the higher scale of the CDI (Words and Sentences) is used, the vocabulary levels of the DS group increase. This is due to the higher number of possible words to assess and the inclusion of words a large number of DS infants know, namely food items. If a larger sample of infants could be assessed over a broader age range, then the course of language acquisition could be followed more systematically. The emergence of different word types found in the lexicon of WS and DS infants could be described and the infant followed longitudinally until they were able to combine words, using the Words and Sentences scale.

Given that the infants were able to participate successfully in a preferential looking paradigm, further research using this tool should be very fruitful. This paradigm can be employed effectively to pull apart the various contributions of different factors to word learning. Given that older children and adults with WS seem to learn words under a different set of constraints (Stevens & Karmiloff-Smith 1997), it would be interesting to examine the factors that influence their word learning much earlier in development by manipulating different aspects of the visual or aural stimuli. For example, the whole object constraint could be investigated by saying a novel word to the child while presenting a novel object on one screen and part of that object on the other. If the child abides by the whole object constraint then they should assume the

word labels the whole object and look longer at it. This line of research will be discussed in greater depth in the final chapter of the thesis.

Chapter Summary

The three studies discussed in this chapter highlight the importance of characterising the language abilities of both atypically and typically developing infants as early as possible in development. Parental reports of comprehension showed that the DS group was delayed in language acquisition but was following a normal developmental pathway. Their marked difficulties with production, however, may be an early indicator of the impairments in productive language seen in adults with DS. In contrast, despite reasonably good language when compared to mental age in adulthood, the WS data point to an atypical language profile in infancy. The pattern of comprehension and production in the WS data does not fit with that seen in either the CA-matched or MA-matched groups. Data from the experimental study also show how important it is to consider abilities both in infancy as well as in the steady state. Infants with WS and DS are equally able to map a verbal label on to a picture in a preferential looking paradigm, despite the discrepancy in their language abilities in adulthood. Both clinical groups are equally delayed with respect to the CA controls. These results could not have been inferred from what is known about the mature phenotype, as is also made clear in the Chapter 5.

The following chapter presents three studies of lexical and syntactic comprehension which were carried out with older children and adults. This was done so that the mature phenotype could be verified and compared against language performance in infancy.

5

Specific profile: Language understanding in older children and adults with Williams Syndrome and Down's Syndrome

Introduction

This chapter is concerned with the receptive language ability of older children and adults with Williams Syndrome and Down's Syndrome. The first part of the chapter gives an overview of the previous research into language ability in WS and DS individuals. In addition to the WS and DS research, patterns of behaviour seen in the normal steady state are mentioned in the appropriate places. Given that the main body of the empirical section of this chapter concerns syntax, this overview concentrates on syntactic understanding but attainments in vocabulary are also referred to briefly.

Part One: Literature Review

5.1 Williams Syndrome

Earlier research with individuals with WS suggested that their language was intact and a great deal better than would be expected for their mental age. However, more recent studies have indicated that this view is too simplistic. In fact, although language is a strength relative to non-verbal abilities and often seems quite impressive, there are several abnormalities in the way these individuals use and understand language. These abnormalities are particularly apparent in aspects of

morphology and syntax. This overview will touch briefly upon research dealing with the receptive lexicon, but its main focus will be on syntax and morphology.

5.1.1 Lexical comprehension

Relative to their non-verbal ability, individuals with WS have quite large and varied vocabularies (Bellugi et al., 1988; Rossen et al., 1996; Stevens, 1996). However, this does not mean that their vocabulary abilities are normal. While they do well on receptive vocabulary tasks such as the BPVS and its American equivalent, the PPVT, they still perform below chronological age level and exhibit other abnormalities. These include difficulty with producing definitions (Bellugi et al., 1988) and less sensitivity to word frequency (Rossen et al., 1996; Karmiloff-Smith et al., 1997). For example, individuals with WS in the Rossen et al. study used a secondary meaning of a homonym, such as Bank-Money/ Bank-River more frequently than normal 10 year olds or MA-matched controls with DS.

As already described in Chapter 4, it has been suggested that the lexicon is acquired differently in WS. Stevens and Karmiloff-Smith (1997) found that while individuals with WS employed both fast-mapping and the mutual exclusivity constraint when learning new words, they did not abide by the whole object constraint or exhibit the taxonomic bias. Typically developing three-year-olds employ all these constraints in word learning.

The absence of the use of certain constraints suggests that the WS lexicon may be semantically weaker than the normal one. In support of this, data from ERP studies suggest that semantic processing in sentences is atypical in this group WS (Neville, Mills & Bellugi, 1994). For example, adults with WS do not process sentence meaning in the normal way. In one study, WS adults and children (from 8 years old) were presented with sentences aurally. Either they ended semantically appropriately, e.g., “I take my coffee with cream and sugar” or inappropriately, e.g., “I take my coffee with cream and radiator”. The WS group showed a large P200 300 response that is not present in normal age-matched controls and an N400 response over the left temporal lobe which was much larger than the response seen in controls. These results

suggest that semantic processing is different in the WS group, but do not tell us how it differs.

However, research investigating single word priming by Tyler et al. (1997) suggests that the semantic structure of the lexicon in long-term memory may be intact. A WS group showed the same priming effects as controls for both taxonomic relations (table-chair) and thematic relations (broom-floor). The fact that individuals with WS exhibit semantic priming for words suggests that the ERP results cited above actually point to a difficulty in integrating semantic information in sentences. This problem with integration is also apparent in the grammatical processing tasks which are discussed below.

5.1.2 Grammatical comprehension

The studies presented in this chapter concentrate on syntactic understanding, but previous research has also been carried out to examine morphology.

Morphology

The most widely studied aspect of morphology in WS is past tense marking. Studies have produced mixed results. Clahsen and Almazan (1998) present data which suggest that individuals with WS have problems with irregular past tense morphology. They exhibit a tendency to over-regularise irregular past tense verbs. So, when presented with a novel verb which rhymes with an existing irregular verb, they will add -ed when one would expect an irregular response, e.g., /sleep/ /slept/ so /shreep/ /shrept/. The authors of these studies argue that these data suggest that past tense formation is a product of two mechanisms: one for regular forms and one for irregular forms. They suggest that in WS the -ed rule system is intact and the other system is impaired. However, other data from a more recent study by Thomas et al. (submitted) on a much larger WS sample suggest that in fact when verbal mental age is controlled for, individuals with WS show no selective deficit in irregular past tense performance, but instead behave like younger, typically-developing controls. It may be that

language delay in the WS group is giving rise to the seeming dissociation responses in the Clahsen and Almazan study.

Individuals with WS also have trouble with the assignment of grammatical gender. This is an aspect of morphology which comes naturally to typically-developing French-speaking children for instance, but causes second language learners problems (Karmiloff-Smith, 1979). In an elicitation study, Karmiloff-Smith et al. (1997) found that individuals with WS had trouble assigning the appropriate gender markings to adjectives when nonce terms were used. So, if a participant was asked to add an adjective to a feminine sounding nonce term, e.g. /podine/ then one would expect the adjective to be put into the feminine form. The results of the study suggest that participants can assign gender to existing words, but that they are unable to operate on several elements of a sentence at once, maintaining gender concordance across a phrase. This suggests they have difficulty in integrating the elements of the language. The individuals were able to use associative learning to match an adjective to a nonce term but were not able to apply rules over multiple parts of a sentence. This pattern of weak extraction of rules and poor system building is also seen in other aspects of cognitive functioning in WS (Johnson & Carey, 1996).

Syntax

Several studies that examine the understanding of syntax have been conducted with WS individuals. These use either on-line (implicit) tasks or off-line (explicit) tasks and highlight the importance of both measures. Karmiloff-Smith et al. (1998) found that individuals with WS displayed a different pattern of abilities when tested using explicit measures of language understanding (picture matching) than when tested using an implicit task (word monitoring). When older WS individuals aged from 14;9 to 34;8 were asked to respond to a particular word in a sentence, Karmiloff-Smith et al. found that, for some types of sentence, response latencies varied according to whether the sentence in which the word appeared was grammatical or non-grammatical. This is the pattern that one would expect for normal controls (Marslen-Wilson, Brown & Tyler, 1988; Tyler, 1992). However, unlike the control group, the

individuals with WS were not sensitive to all of the grammatical violations presented. While they were able to detect violations of auxiliary verbs (e.g., /She **was have** breakfast/ instead of /She **should have** breakfast/) and phrase structure (word order), with increased response latencies to target words in an ungrammatical sentence, they did not appear to detect violations of subcategory structure. So, response time to a target in a sentence such as /Mary **listened** the teacher/ was no longer than to /Alice **listened to** the radio/. The authors suggest that individuals with WS can understand the subcategory structure but that they are slow to integrate the information into their processing of the whole sentence. This is reflected in the equally long response times to subcategory sentences, whether correct or incorrect. It is likely that these structures are processed lexically, so a word such as /listen/ would activate various potential argument structures. The word then has to be integrated into one of these potential structures. It is the speed of this integration that may be compromised in WS.

Explicit tasks yield different results, due to their greater meta-cognitive demands. The majority of these tasks involve the participant choosing the one picture, amongst several distractors, which corresponds to the meaning of the sentence presented to them. In these tasks participants must decode the speech of the experimenter, remember the sentence, process the picture and then pick the one that matches the sentence. These demands increase the difficulty of the task for individuals with general cognitive impairment, such as those with WS. Although individuals with WS perform quite well on similar tasks with single words, such as the BPVS, it should be noted that these entail less cognitive demands (Karmiloff-Smith et al., 1997). In particular, they do not require the participant to remember or process whole sentences.

Early studies of syntactic understanding in WS, using very small groups and subsets of standardised tests such as the TROG (Test of Reception of Grammar), indicated the individuals with WS had a good understanding of syntax. Bellugi, Bihrlé, Jernigan, Trauner and Doherty (1990) report data from 3 participants who performed well on a subset of items including those tapping the understanding of passives, a linguistic structure that is difficult for young typically-developing children and on which MA-matched individuals with DS do poorly. However, this was not a full assessment of

syntax. In order to address this problem, Karmiloff-Smith et al. (1997) collected data from the full version of the TROG from 18 participants aged 8 - 34 years old (mean 18;2 years). They found that individuals with WS perform at a level well below their chronological age, reaching a mean test age of only 6;3 years. This is similar to their performance on a non-verbal task, Ravens Progressive Matrices. The WS group has particular difficulty with sentences which have embedded structures, e.g. The boy the dog chases is big. Such sentences place large demands on language processing because the complex linguistic structure must be decomposed in order to retrieve its meaning. These difficulties with grammar are not confined to the English language. They have also been found in Italian participants with WS (Volterra, Capirci, Pezzini, Sabbadini & Vicari, 1996).

In addition to data from the TROG, a further study using a similar procedure but tests of more complicated structures, such as active and passive sentences with agentive and non-agentive verbs, found that individuals with WS had great difficulty with these structures (Karmiloff-Smith et al., 1998). Their performance was much worse than that of normal controls, and more like elderly controls who had left school at 14. Both the groups had problems with similar structures, especially those with non-agentive verbs and deverbal adjectives. Both the elderly and WS groups found sentences with locatives and agentive verbs easiest. This suggests that the WS group are behaving like less educated controls. Interestingly, the elderly group behaved like normal controls, and quite unlike the WS group, on the subcategory constraint task. This result suggests that there is likely to be some aspect of language processing in WS which is very different from normal and represents, perhaps, a language-specific impairment not merely due to less education or to general cognitive impairment.

What, then, are the causes of the pattern of strengths and weaknesses within language in WS? Meta-cognitive skills certainly have a role to play in language ability and these are rather more important for off-line tasks than on-line tasks. Individuals with WS are likely to have meta-cognitive impairments due to their general cognitive impairments. They perform better on tasks which make lower cognitive demands,

such as vocabulary tests like the BPVS, than those which are more demanding such as sentence-picture matching which assesses aspects of syntax.

The ability to extract rules and generalise them also has a differential effect on aspects of language. It is known that individuals have a problem extracting the rules which govern gender marking in French, for example (Karmiloff-Smith et al., 1997). The construction of the lexicon, a relative strength in WS, relies less on the ability to generate a rule-based system, than might the correct use of syntax, which is impaired. Related to this, is the relative strength of phonological short-term memory in WS. Because individuals with WS are able to learn items in the language by rote and perhaps devote more representational space to language, they are less inclined to extract regularities in the system and generalise them to new situations. This is evident in non-word repetition performance, where people with WS never reach the level of typically developing 6 year olds, who start to link the novel words to existing entries in their lexicon, in order to bootstrap the representations.

There is also increasing evidence that individuals with WS rely heavily on phonology in language, and relatively less on semantics (Karmiloff-Smith, Brown, Grice & Paterson (submitted). For example, in a word monitoring task, participants' inability to detect subcategory violations may have been due to the fact that semantic information was not available quickly enough to be integrated into the sentence (Karmiloff-Smith et al., 1997). It has also been found that in reading, participants with WS have difficulty forming a semantic representation of single words. Vicari and his colleagues found that learning of auditorily presented words was improved if a picture of the item was presented alongside the auditory stimulus, to provide the semantic code for the word (Vicari, Bellucci & Carlesimo, manuscript). In addition, Laing, Hulme and Karmiloff-Smith (submitted) found that their participants with WS read concrete and abstract words equally well, whereas children in a control group found concrete words, which were imageable, much easier to read. This suggests that the semantic representations in WS may be shallow and that individuals with WS may rely more on phonology to form representations of language. This has a differential effect on certain aspects of language. Semantics are crucial when trying to process

language on a syntactic level, in sentences, but may be less so for operating at the lexical level. For example, in a sentence when there are two protagonists, e.g. “The cat hits the dog”, one has to be clear of the meaning intended in order to decode the word order. However, a clear phonological representation and a weak semantic code may be sufficient to operate at the word level.

5.1.3 Summary

There is no doubt that some aspects of WS language are better than one would expect given non-verbal mental age. The vocabulary levels of individuals with WS are better than their MA, but not at the level of their CA, and they produce longer and more complex utterances than one would expect given their non-verbal mental age (Reilly, Klima & Bellugi, 1990; Volterra et al., 1996). Despite this, there are several aspects of language which seem more seriously impaired, many of which are yet to be investigated. Certain aspects of syntax, such as subcategory constraints and embedded clauses, cause problems for individuals with WS. Their problems with these structures are not like those of poorly educated controls, but are probably of a different nature. In addition to these syntactic difficulties, they also have problems with aspects of morphology, such as gender marking (Volterra et al., 1996). It is in the fine analysis of language that one finds deficits in WS language production and processing, suggesting that further systematic and thorough investigations of other aspects of language should be carried out. Certainly, it is incorrect to characterise language in WS as “intact”.

5.2 Down’s Syndrome

Research suggests that individuals with DS have many problems with language. This group is certainly thought to perform at a lower level on language tasks than MA-matched individuals with WS (Singer Harris et al., 1997). The following section is a brief overview of research on specific areas of DS language. The areas covered are vocabulary comprehension and syntactic comprehension. These were chosen because there is complementary literature also available for WS.

5.2.1 Lexical comprehension

Several researchers have suggested that lexical processing is better than syntactic processing in DS, especially in later stages of development (Fowler, 1990; Chapman, Schwartz, & Kay-Raining Bird, 1991; Rosin, Swift, Bless & Vetter, 1988). Certainly, individuals with DS perform at their mental age level on vocabulary tests. Rosin et al. (1988) found that adolescent boys with DS performed as well as mental age matched controls on the PPVT, a test of receptive vocabulary. It was also found that on a test of receptive grammar, called the TACL, individuals with DS performed much better on the first part, which is reliant on lexical comprehension and taps vocabulary and semantic relations, than on the later 2 parts (Chapman et al., 1991). However, as discussed below, the pattern of MA-level performance on lexical tasks is not found for grammatical understanding.

5.2.2 Grammatical comprehension

Morphology and syntax

Individuals with DS have more difficulty with morphology and syntax than with single words. Their syntactic comprehension ability lags behind that of MA-matched children (Miller, 1988; Rosin et al., 1988; Chapman et al., 1991). Chapman and her colleagues (1991) found no difference between DS children's scores on the Peabody Picture Vocabulary Test, or on the TACL-R (a test of syntactic understanding) when compared to those of MA-matched children. However, when the two tests were compared, the difference between lexical and syntactic performance was more marked in the DS group. On the TACL-R test, the individuals with DS (aged 5 –20 years) had much more difficulty with grammatical morphemes and with elaborated sentences and reversibles subtests than with word classes and relations. The subtests with which they had difficulty assess aspects of grammar such as negation, reversible sentences and active and passive structures. Marcell, Cohen and Sewell (1990) report similar results. Adolescents and adults with DS in their sample perform as well as controls with mental retardation, matched on IQ and chronological age, on a vocabulary test. However, the DS group has more problems than the control group

with items on the Miller-Yoder Test tapping singular/plural, possessives, reflexives, verb inflections and passives. This suggests that individuals with DS have particular problems with morpho-syntax which go beyond their general cognitive impairment.

Other tests have also highlighted the DS grammatical difficulties. Vicari, Caselli and Tonucci (2000) have found that younger children with DS (4 -7 years old) perform poorly on repetition tasks which assess the processing of morphology and syntax. The children with DS make numerous errors, especially with function words, which are an important building block for grammar.

The grammatical difficulties experienced by individuals with DS could arise from their propensity to focus on different aspects of the language than those focused on by normal adults. Fowler (1984) and Bridges and Smith (1984) suggest that, when decoding word order, individuals with DS place far more emphasis on semantics than on the syntactic structure of the language. Individuals with DS will choose pictures which are semantically plausible over those which match the syntax of a sentence. In addition, Naigles, Fowler & Helm (1992) found that school-aged children with DS also interpreted ungrammatical sentences in a different manner from controls. When the children were presented with sentences in which ungrammatical verb frames were used, e.g. /the zebra goes the lion/ normal controls make a verb-based interpretation. That is, when given a toy zebra and lion, they move the zebra towards the lion. On the other hand, children with DS are likely to base their interpretation on the sentence frame, so make the zebra push the lion. This reliance on semantics by individuals with DS may cause difficulty when meaning cannot provide support for comprehension, for example, when the meaning of the sentence is unusual, as in the Naigles et al. study.

Why, then, is there such a discrepancy between lexical and syntactic understanding in DS? Several reasons have been suggested for this. These include: hearing problems, short-term memory deficits and general cognitive impairment, all of which were discussed in greater depth in Chapter 4.

5.2.3 Summary

Lexical comprehension in DS appears to be at a level equivalent to mental age, whereas aspects of syntactic comprehension are rather more impaired, well below the mental age level. Individuals with DS have difficulty on syntactic tasks which involve structures such as passive sentences and reversibles. They also have problems with morphology, such as grammatical gender marking and verb inflection. It is likely that the discrepancy between lexical and syntactic performance may be due to a number of factors interacting with one another. Possible causes of syntactic difference might include problems with hearing, short-term memory or the propensity to focus on semantics rather than syntax when trying to process syntactic structures.

5.3 Literature Summary

This overview points to a difference between lexical and syntactic understanding in DS which is greater than that reported for WS, although both groups have particular difficulty with syntax and morphology. Some striking similarities and differences in the possible causes of the impairment emerge from the overview of the WS and DS literature. Whereas there seems to be an over-reliance on phonology and rather weak semantic coding in the WS case, the DS group appears to exhibit the opposite pattern. In addition, where the DS group have impaired phonological short-term memory which might be a contributor to language problems, the WS group have relatively good phonological short-term memory. This is interesting because it suggests that difficulties with syntax in the two groups might stem from different factors. Of course, both groups have common impairments too. General cognitive impairment has a role to play in reduced meta-cognitive skill, which is important for explicit language tasks, and both groups are likely to take longer to integrate the syntactic elements of a sentence, due to slower processing.

Now that an overview of lexical and morpho-syntactic comprehension has been provided for both WS and DS, the empirical work can be introduced. The following studies were conducted to investigate the understanding of two aspects of syntax in

groups of older children and adults with DS and WS. They will enable a much-needed systematic comparison of language ability across the two groups.

Part Two: Empirical Studies

5.4 Introduction

This section is divided into three parts. The first concerns vocabulary ability in older children and adults with WS, DS and their MA and CA-matches. This will enable a test of the hypothesis that WS vocabulary is better than MA-level, which has been suggested in previous research (Bellugi et al., 1988). The other two parts involve the assessment of the understanding of two aspects of syntax which have been assessed in separate studies with adults with WS but have not been compared across groups. These are word order and subcategorisation. They will be assessed using new elicited imitation tasks, in which participants are asked to repeat sentences which were either correct or incorrect grammatically. It was predicted that sensitivity to word order violations would be high in the WS group, as reported in previous research (Karmiloff-Smith et al., 1998), but that the DS group would not be sensitive to such violations. This might occur because individuals with DS are known to perform below MA level on tests of relatively simple syntactic understanding, such as the TROG, whereas individuals with WS perform at around MA level (Karmiloff-Smith et al., 1997). By contrast, it was hypothesised that the DS and WS groups would both have difficulty detecting subcategorisation violations. The WS impairment has been reported in previous research and is expected to be shared by the DS group, because the task is more demanding than more common tests of syntax (Karmiloff-Smith et al., 1998). It is likely that the WS group would exhibit higher levels of language comprehension than the DS group, given the WS cognitive profile, but that their language comprehension would not be normal. Characterisation of the lexical system in WS has shown that the lexicon in WS behaves atypically (Bellugi et al., 1988; Stevens & Karmiloff-Smith, 1997) and this pattern could also be found in the syntactic system.

5.5 Experiment One: Lexical understanding - Performance on the British Picture Vocabulary Scale

Given the claims about the high level of receptive vocabulary scores achieved by individuals with WS, a study was conducted to ascertain whether the receptive vocabulary of this group was as good as is claimed. This was done by making a direct comparison between individuals with WS and those with DS matched pair-wise on mental age from the BAS II and chronological age.

5.5.1 Method

Participants

7 individuals with WS and 7 individuals with DS, who were CA and MA-matched to individuals in the WS group were tested. Their chronological ages and mental ages are given below.

Table 5.1 Chronological and mental ages for each group

	Mean CA (years)	range	Mean MA (years)	Range
WS	21;6	9;10-33;4	6;9	5;1-9;4
DS	22	9;11-33;7	5;9	5;1-6;4

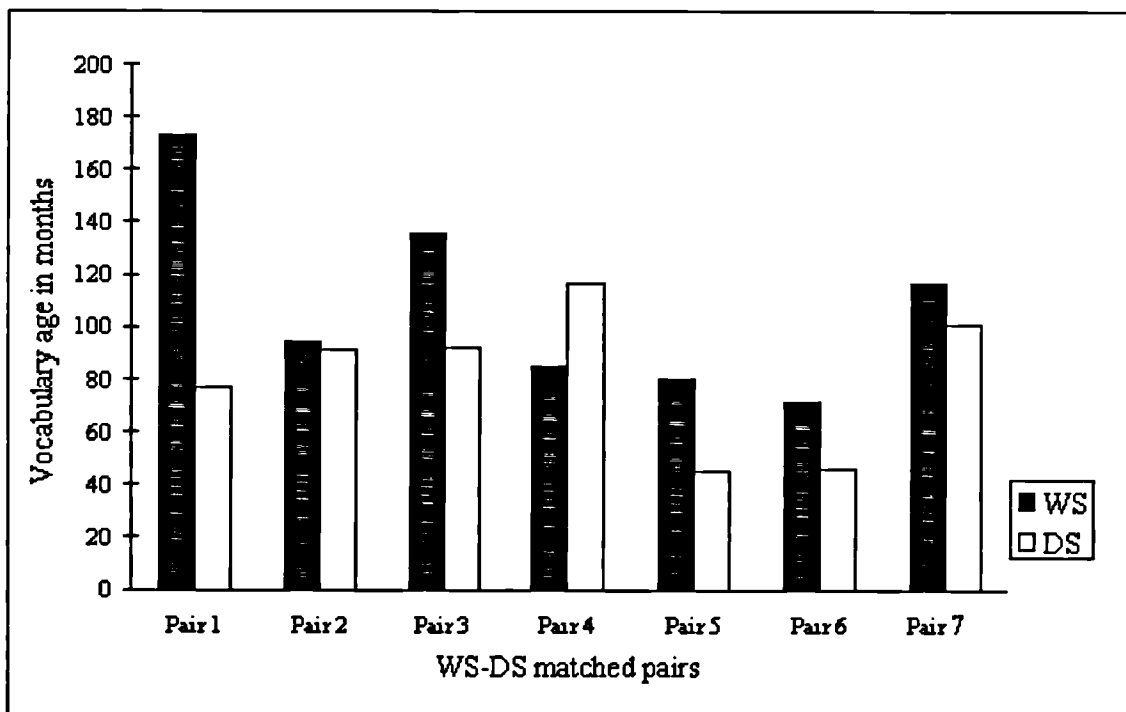
Procedure

All participants were tested using the British Picture Vocabulary Scale (BPVS). In this test, individuals are asked to point to 1 of 4 pictures which corresponds to a word they hear. It assesses single word comprehension. The words are increasingly infrequent and abstract as the test proceeds. Testing took place in a quiet room as part of a longer testing session, and was conducted in accordance with the standardised procedure in the manual.

5.5.2 Results and discussion

The raw score for each individual was converted into a test age (TA), according to the manual. As shown in Figure 5.1, in 5 of the 7 pairs of participants, the participant with WS had a higher vocabulary age than the participant with DS. This was reversed for one pair (pair 4). A further pair (pair 2) exhibited a negligible difference. A paired t-test indicated that there was no significant difference between the mean vocabulary age for the WS group and the DS group, $t(df\ 6) = 1.76, p < .062$. However, this difference approaches significance. Further research involving larger groups might uncover a significant difference. (With significance level of 95%, a power of 80% on the observed differences would require 17 WS/DS pairs. The small sample size gives a large variance due to pair 4, thus resulting in a rather conservative power calculation.) Overall, it appears that most of the individuals with WS have a higher vocabulary age than their counterparts with DS.

Figure 5.1 Differences in vocabulary age for WS-DS matched pairs



A further analysis was carried out to investigate the difference in the discrepancy between chronological age and vocabulary test age for the WS and DS groups. The difference between chronological age and test age was calculated for each participant.

A paired t-test revealed that there was a greater discrepancy between CA and TA for the DS group than the WS group, $t(df\ 6) = 2.33, p < .05$. This was as expected, given the reports of relatively spared vocabulary in WS (Bellugi et al., 1988). This method of analysis allowed the differences within individuals to be compared, so that vocabulary levels could be assessed relative to chronological age. This is important because it highlights the relative sparing of vocabulary and the fact that vocabulary is not at chronological age level in either WS or DS. Both groups are performing at a level below their chronological age.

5.6 Experiment two: Sensitivity to word order and subcategory constraint violations.

5.6.1 Introduction

This experiment examined sensitivity to grammatical violations using a qualitative analysis of responses. It was predicted that the WS group would show more sensitivity to word order violations than the DS group and would behave like normal controls, but that the WS and DS groups would both experience difficulties with subcategory constraint violations.

5.6.2 Method

Participants

8 individuals with WS, 7 individuals with DS, CA and MA- matched to the range of the WS group, 8 typically developing children with CAs which match the mental age of the WS group (MA-matched group) and 8 individuals CA-matched to the WS group were tested. However, only 5 individuals with DS could perform the task. Also, only 7 MA-matches could perform the task. The mean CAs and MAs for the participants whose results were included in the analysis are given below. These matches were not as close as would be ideal due to limited numbers of DS participants.

Table 5.2 Chronological and mental ages for each group

	Mean CA (years)	range	Mean MA (years)	Range
WS	20;9	10;11-32;9	6;9	5;1-9;4
DS	26;3	17;11-35;3	5;11	5;7-6;5
MA	6;11	5;2-8;11	-	-
CA	19;10	9;10-29;8	-	-

Stimuli

Subcategorisation frames

The stimuli were 19 pairs of sentences that differed in length. In one sentence of each pair, the subcategorisation constraints on the verb were not violated; in the other these constraints were violated. There were two types of sentence. The first type was correct when a preposition came after the verb (1). The second type was correct if there was no preposition after the verb (2). If participants are sensitive to the violation, they should take longer to respond to the incorrect sentences.

- | | | |
|---|---------------|---|
| 1 | Grammatical | I never listen to the radio |
| | Ungrammatical | I always listen the teacher |
| 2 | Grammatical | I always hear my mother when she calls me |
| | Ungrammatical | I always hear to my sister when she cries |

A short version of the sentences in this task was also devised which kept the subcategorisation frame part of the sentence, but shortened other parts of the sentences. This was used when the memory load of the original version was too high for a participant.

Word order

Ten pairs of sentences were generated. One in each pair had the correct word order (1), the other had a word order violation (2). If participants are sensitive to the

importance of word order for syntactic coherence, then they should take longer to respond to the ungrammatical sentences.

- 1 I borrow three books from the library every week
- 2 I borrow four books from library the every month

A short version of this task was also prepared, as for subcategorisation frames described above.

Examples of both word order and subcategorisation sentences are provided in the Appendix.

Procedure

The participants listened to recordings of all the stimuli through headphones. They were asked to repeat what they heard (“You say what the lady said”) immediately after the sentence ended. This instruction was given at the beginning of the task. Their responses were recorded using a DAT recorder. They were transcribed later. The response latency of the participants, that is the time between the end of the recorded sentence and the beginning of the response, was also recorded. A practice tape was used to familiarise the WS, DS and MA-matched group with the task. The CA- matched group was simply familiarised using the first two sentences of the test. These were neutral and not included in the analysis.

5.7 Results

As described in the procedure section, two types of data were gathered: transcripts of the participants’ responses and response times in milliseconds. The qualitative data was of particular interest for this thesis because response types were not considered in the previous work on sensitivity to violations of word order and subcategorisation frames. This was the principal analysis chosen because it may highlight subtle differences between the WS and DS groups which might be missed by the response time data, due to huge variability and the fact that both groups exhibit slow reaction

times. The second analysis was a comparison of different response times across the four groups of participants, but these data will not be fully discussed for the above reasons.

The results of the present study turn out to be like those found using explicit tasks in which participants had to point to a picture which matched a sentence read by the experimenter. It was hoped that the procedure employed in the present study could be used as an alternative to the lengthy word-monitoring procedure used by Karmiloff-Smith et al. (1998). In that task, participants with WS found it difficult to attend to the word-monitoring task for long periods and appeared to miss the inter-personal contact normally present in testing situations. It was thought that the more interactive nature of a sentence repetition task would enable the collection of additional data, while keeping the participants happy. Unfortunately, the reaction time results from the repetition task were not as sensitive as those from the word-monitoring task. It is likely that the memory demands of the task clouded its sensitivity to small changes in language processing. This will be discussed further in section 5.9.

5.7.1 Response content

The types of responses given to grammatical and ungrammatical sentences were considered. This analysis included both errors and correct responses. It was important to consider the qualitative nature of responses because the participants may have interpreted the task in different ways. Several of the participants produced verbatim responses when presented with an ungrammatical sentence. It is difficult to interpret these responses because if someone is asked to repeat the sentence they have heard, then both a verbatim repetition and a spontaneous correction are equally acceptable. It was hoped that a more detailed analysis of responses might highlight interesting patterns across ungrammatical and grammatical sentences.

Each response was coded into one of the following categories.

Acceptable:

Verbatim or close to verbatim response which preserves meaning.

Responses of this sort were deemed acceptable because they indicate that the participant has preserved the main elements of the sentence. In addition, the changes are so small that they are likely to be spontaneous and not due to the grammatical violation. Acceptable responses are also allowable in the case of ungrammatical sentences. This is because they conform to the task demands to the same extent as responses involving changes.

Grammatical change to critical part:

The part of the sentence being examined is changed, but the result is grammatical, e.g.

The doctor told him to eat less fat. (Model: The doctor advised him to eat less fat).

The doctor told him to eat less fat. (Model: The doctor advised to him to eat less fat).

The critical part of the sentence is where the violation takes place in the incorrect model. A grammatical change in the response is a change which is correct but different from that expected when the task was designed. This was developed as a category because several types of change to ungrammatical models are equally acceptable.

Ungrammatical change to critical part:

The part of the sentence being examined is changed, but the result is ungrammatical, e.g. *He arrived to the airport just in time to meet his son.* (Model: He arrived at the airport just in time to meet his son.) or

He arrived to the airport just in time to meet his son. (Model: He arrived the airport just in time to meet his son.)

The critical part of the sentence may be changed incorrectly, either in grammatical sentences or ungrammatical sentences. In the latter case, this may occur when the participant has difficulty coping with the processing demands caused by a grammatical violation.

Other:

A response which does not preserve the meaning of the original sentence or may be only a fragment, e.g. *The girl take a big breath enters the big room.* (Model: The nervous girl took a deep breath before she entered the room.)

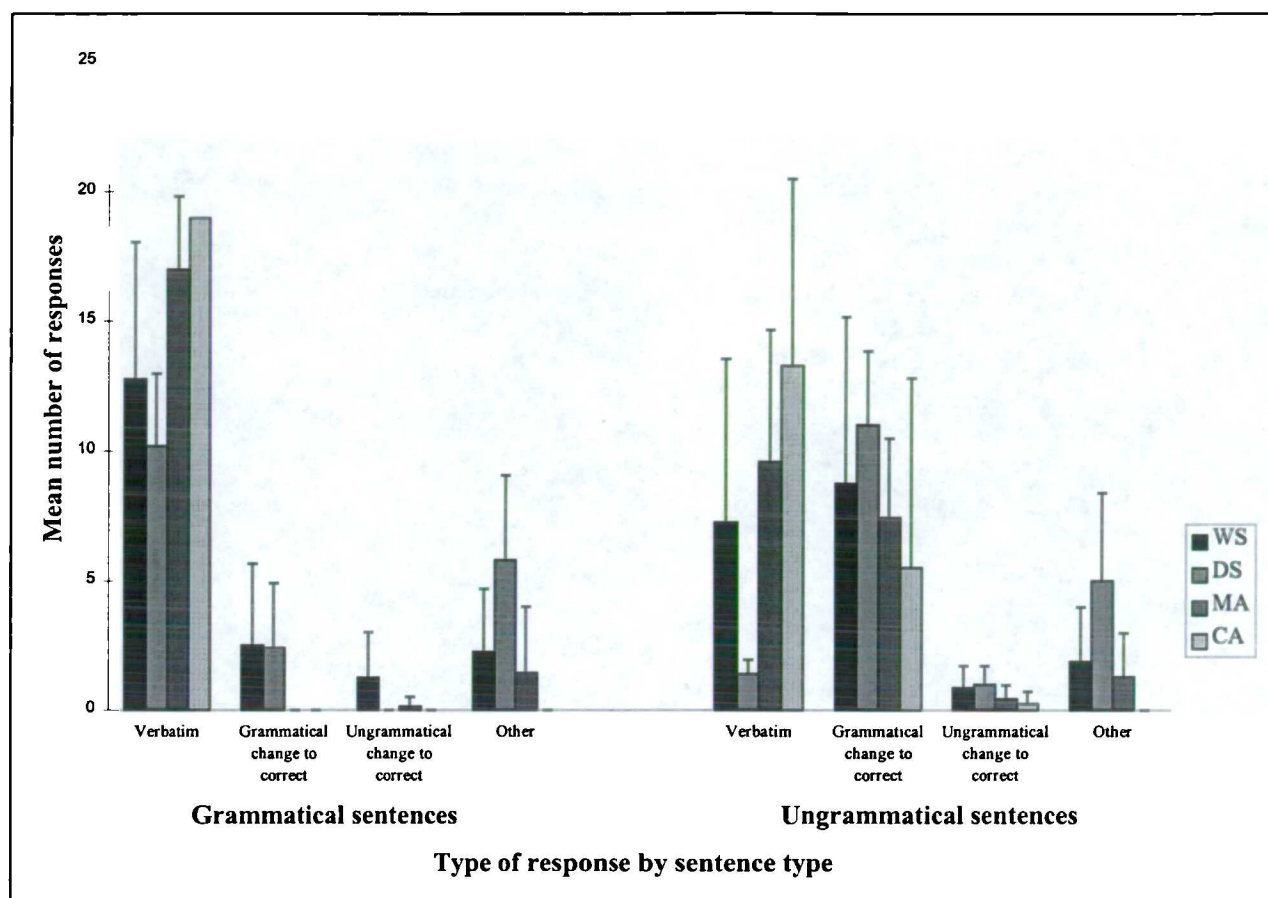
This suggests that the participant was unable to remember all the material or was unable to produce such a complicated utterance.

The number of responses of each type was recorded for each participant. These were then transformed into percentages of the total responses, out of 19 for subcategorisation sentences and out of 10 for word order sentences. This was done so that responses across the four response types could be compared effectively. The proportion of responses of each type was then compared across groups to examine whether group differences were present. A repeated measures Anova, with type of sentence (grammatical versus ungrammatical) and response type (acceptable, grammatical change, ungrammatical change and other) as the within-subject factors and group as the between-subjects factor was carried out. This was done separately for the subcategorisation and word order sentences. All these analyses were carried using the more conservative Greenhouse-Geisser F test because of the heterogeneity of the data.

5.7.2 Subcategorisation results and discussion

The results for subcategorisation sentences are presented in Figure 5.2. Readers interested in data from individual participants can find this tabulated in Appendix E. An interaction between response type and group was revealed, $F(4.79, 38.29) = 7.05$, $p < .001$. In addition, there was an interaction between sentence type and response type, $F(1.09, 26.19) = 32.28$, $p < .001$ and an effect of response type $F(1.60, 38.29) = 72.70$, $p < .001$. No effect of sentence type or group was found and there were no further significant interactions.

Figure 5.2 Mean number of responses by sentence type for subcategorisation



Simple main effects showed that the interaction between response type and group arose from several differences between groups. There was a difference between groups for verbatim responses to grammatical sentences, $F(3,27) = 9.00$, $p < .001$. Post hoc tests revealed that the difference lay between the WS and the CA-matched group (Tukey HSD $p < .05$) and the DS group and both CA and MA controls. The CA-matched controls produced more acceptable repetitions of grammatical sentences than either the WS or DS groups. The MA group was also better than the DS group. A difference was also revealed for “other” responses to grammatical sentences, $F(3,27) = 6.90$, $p < .005$, with the DS group producing significantly more “other” responses than the CA and MA groups, Tukey HSD, $p < .05$. For comparison with WS this difference was approaching significance, $p < .052$. (Assuming equal variance in the WS and DS groups, in order to achieve a significance level of 95% with a power of 80%, 13 participants in each group would need to be tested.) Another simple main

effect was found for acceptable responses to ungrammatical sentence, $F(3,27) = 4.54$, $p < .05$. Post hoc tests indicated that the difference lay between the DS and CA groups, with the DS group producing fewer verbatim responses to ungrammatical sentences than the CA group. The final significant difference between groups was found for “other” responses to ungrammatical sentences, $F(3,27) = 6.70$, $p < .005$. Post hoc tests revealed the difference lay between the DS group and all other groups, Tukey HSD, $p < .05$, with the DS group producing significantly more “other” responses. Post hoc t tests revealed that the interaction between sentence type and response type resided in the difference in the proportion of acceptable and correct responses to grammatical and ungrammatical sentences. There was a significant difference in the proportion of acceptable responses to grammatical and ungrammatical sentences, $t(df\ 27) = 5.88$, $p < .001$, with more acceptable responses to grammatical sentences (the optimal response) than to ungrammatical sentences. The opposite effect is seen for the proportion of grammatical changes to the critical part of the sentence, $t(df\ 27) = -5.99$, $p < .001$, with more changes to ungrammatical sentences.

Overall, the participants were behaving as one would expect, making more changes to the critical part of ungrammatical sentences than to grammatical sentences. This suggests that they are detecting the errors and changing them. They were also making a larger number of acceptable responses to grammatical sentences, that is, repeating the sentences exactly as heard or with minor changes.

In more detail, the CA group produced significantly larger proportion of acceptable responses to grammatical sentences than the WS and DS groups. In addition, the DS group’s performance was poorer than that of MA controls. These findings suggest that both atypical groups had difficulty in repeating the sentences correctly. There are two possible reasons for this. The first is that the syntactic demands of the sentences were too high for the participants. Even when sentences were correct, it may have been that the processing demands placed upon subjects by relatively complex sentence structure meant that they could not respond appropriately. Individuals may have had difficulty integrating all the grammatical elements into the sentence. The second concerns memory load. Participants may have had difficulty holding the sentences in memory

and could therefore have been unable to repeat the sentences verbatim or to remember the gist. Preliminary analyses suggest that longer sentences (measured in morphemes) gave rise to more “other” responses than shorter sentences (mean for short .020, mean for long .047)³. This occurred for both ungrammatical and grammatical sentences.

For both ungrammatical and grammatical sentences, the DS group produced a significantly higher proportion of “other” responses than the control groups. For ungrammatical sentences, the DS group also produced a significantly higher proportion of “other” responses than the WS group. These results may highlight the particular contribution of memory to the responses of the DS group. This group was unable to make suitable responses, either for those sentences for which only acceptable responses were necessary or for those which were ungrammatical and required some form of change. Individuals with DS have difficulty imitating sentences and generally produce short utterances with a simple structure (Vicari et al., 2000), so these results are not surprising. Whereas the WS group did not produce the same proportion of acceptable responses as the CA controls for grammatical sentences, they did not differ from the controls in the proportion of their “other” responses. The WS group made more changes or acceptable responses to the ungrammatical sentences than their counterparts with DS, which suggests that they are more sensitive to grammatical violations than the DS group.

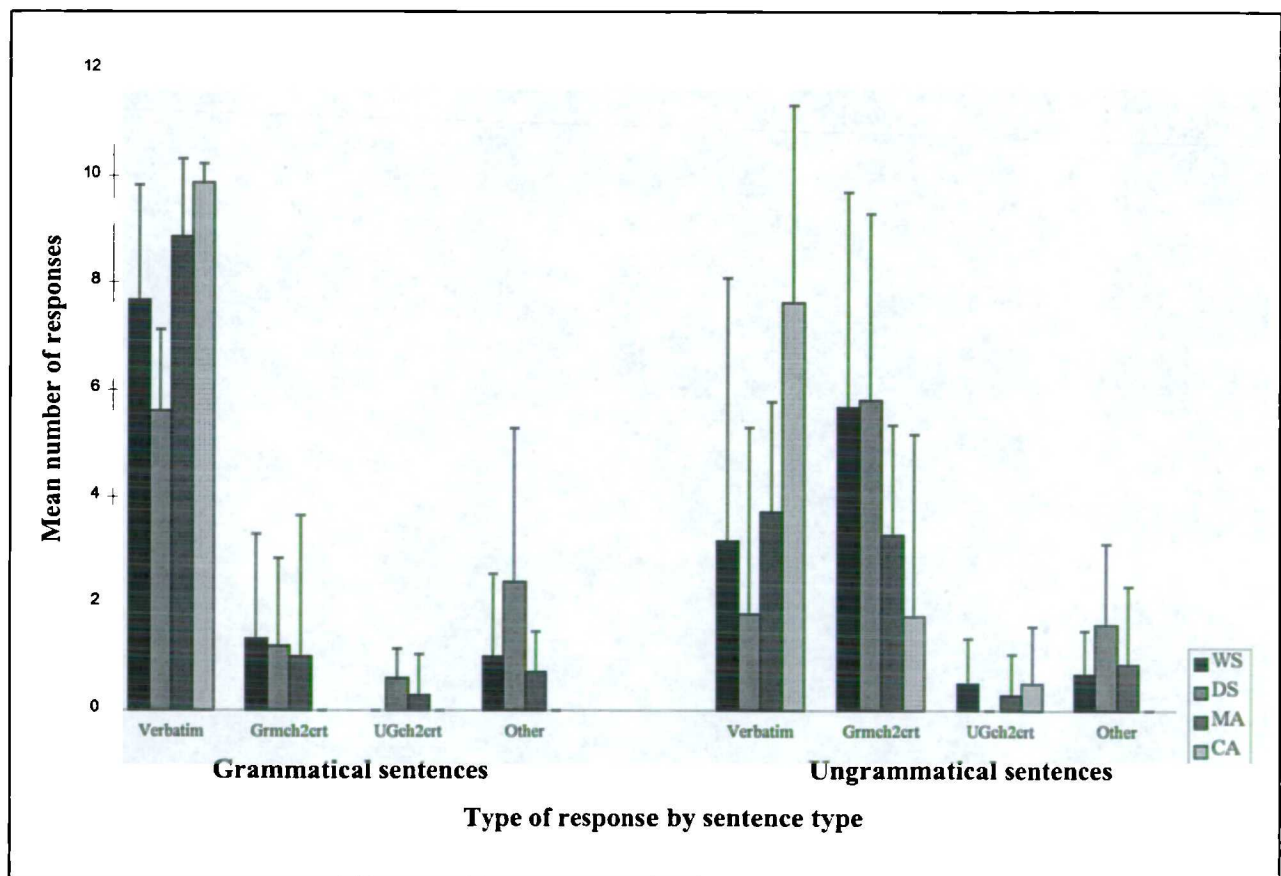
The subtle differences in the responses of the WS and DS groups might be the key to the differences between language ability in individuals with WS and DS. The WS group appears to be retaining larger parts of the sentence in their responses than the DS group and this might point to slightly more efficient language processing. This is particularly noteworthy given the fact that all the participants with DS were given the short version of the task and should therefore have experienced a lesser memory load than most of the participants with WS.

³ The two shortest sentence lengths in morphemes were considered the shorter sentences. The two longest sentence lengths in morphemes were considered the longer sentences, irrespective of which version of the task was used.

5.7.3 Word order results and discussion

The same types of analyses were carried out for the word order experiment and data are presented in Figure 5.3. Readers interested in individual data can find this tabulated in Appendix F. The data were analysed using a repeated measures Anova with sentence type and response type as the within subjects factors and group as the between subjects variable. There was a significant effect of response type, $F(1.62, 35.77) = 57.75$, $p < .001$ and significant interactions between response type and group, $F(4.88, 35.77) = 4.98$, $p < .05$ and sentence type and response type, $F(1.56, 34.32) = 23.37$, $p < .001$. There were no other significant main effects or interactions.

Figure 5.3 Mean number of responses by sentence type for word order



The interaction between type of sentence and type of response was explored further using t tests. These revealed a significant difference in the proportion of acceptable responses to grammatical and ungrammatical sentences, $t(df 27) = 5.88$, $p < .001$, with

more acceptable responses to grammatical than ungrammatical sentences. The opposite is seen for grammatical changes to the critical part of the sentence, where there is also a significant difference in the proportion of such responses for grammatical and ungrammatical sentences, $t(df\ 27) = -5.99, p < .001$. There are more grammatical changes to ungrammatical sentences than to grammatical sentences, as would be expected.

Tests of simple main effects revealed that the interaction between group and response type arose from differences between the groups for three types of response, acceptable responses to grammatical sentences $F(3,27) = 9.01, p < .01$, “other” responses to grammatical sentences $F(3,27) = 6.89, p < .01$ and acceptable responses to ungrammatical sentences, $F(3,27) = 3.26, p < .05$. Post hoc tests revealed that for acceptable responses to grammatical sentences, the DS group produced fewer responses than either the CA or MA groups, Tukey HSD, $p < .05$ and the WS group produced fewer responses than the CA group, Tukey HSD, $p < .05$. There was no significant difference between DS and WS. For “other” responses to grammatical sentences the differences between groups lay between the DS group and the CA group, Tukey HSD, $p < .05$. The DS group produced more “other” responses than the CA control group. For ungrammatical sentences, the DS group made fewer acceptable responses than the CA group, Tukey HSD, $p < .05$.

The data indicate that the DS group produced significantly more “other” responses than the CA control group. Responses in the “other” category indicate that a participant could not make a response which preserved the meaning of the original sentence. In many cases participants giving “other” responses could only reproduce fragments. This point is noteworthy because all members of the DS group were presented with the short version of the task, in an attempt to reduce the memory load, yet still had difficulty in responding correctly. Again, it appears memory ability could be a contributing factor to the impairment in the DS group, with “other” responses increasing with sentence length (mean for shorter sentences .11, mean for longer

sentences .32)⁴. However, as before, it was difficult to separate the effects of memory load and syntactic complexity. Both these factors can be reflected in sentence length.

5.8 General discussion

The present studies investigated the effect of grammatical violations on the type of response given. It appears that individuals with DS have greater difficulty with syntactic processing than individuals with WS, as evident in the data from the subcategorisation task. However, both groups are impaired. The participants with WS produce fewer acceptable responses than the CA group. Further research must be conducted to investigate the cause of these impairments.

5.9 Recommendations for further research

Repetition tasks raise several problems for data interpretation. In the present study, several of the participants, including normal adults, produced verbatim or near verbatim responses when presented with ungrammatical sentences. Although one might expect that if an individual were aware of a grammatical violation, they would spontaneously correct the sentence, an uncorrected response complies as fully to the task instructions as a changed response (or in a sense, even more so). Several participants believed that when they were asked to repeat the sentence, they should repeat it irrespective of whether it was grammatically acceptable or not. It appeared that the response times did reflect sensitivity to violations in normal controls⁵, despite the type of response given, but for the qualitative analysis verbatim responses were harder to interpret. In future studies, participants could be asked to repeat sentences, but to correct any errors if they occurred. In addition, some participants made changes to the critical part of the sentence despite the fact that the original was correct. This may be an indication that the presented sentence does not seem acceptable to them,

⁴ The criteria for shorter and longer sentences were the same as those in note 2.

⁵ Response times to grammatical and ungrammatical sentences for all controls were analysed. One CA control was excluded because response times were not recorded. For the purpose of this test, the data for subcategorisation and word order collated. A significant difference in RTs to grammatical and ungrammatical sentences was found, $t(df\ 27) = 2.679, p < .05$, with means to grammatical and ungrammatical sentences equal to 69.40 secs and 76.91 secs respectively.

but is difficult to interpret. A change to an ungrammatical sentence might differ from the expected response but should nevertheless be deemed correct. There are two main problems with repetition tasks. The first concerns the participants, who may make different interpretations of the task. The second concerns the analysis of responses, in which the underlying reason for a response may be ambiguous. Further research must heed this difficulty.

In addition to examining the receptive language abilities of older children and adults with WS and DS, these studies were also carried out to investigate whether repetition tasks could be used as an alternative to word-monitoring tasks when assessing sensitivity to grammatical violations. This was done because the repetition paradigm works more effectively with individuals with WS than the word-monitoring task. Unfortunately, the data from both the subcategorisation and word order tasks suggest that this paradigm is not sensitive enough to changes in response time to different types of sentence. It appears that the memory load of sentence repetition tasks might be a cause of this problem and slows responses to such an extent that differential effects of sentence type may be lost. The sentences used in this study may be too long for the DS group, as is apparent in the large number of incomplete responses these individuals made. The WS group was able to remember the sentences more clearly but memory or processing demands which were not explicitly controlled for, which were placed on them may have obliterated any effects of sentence type. These problems might be alleviated by using shorter sentences. However, the DS group even had problems with the shorter form of the task and it may not be possible to preserve the structures under investigation in sentences that are much shorter. This difficulty, in itself, reflects the differences in verbal processing between WS and DS.

Further research must be carried out to unravel the causes for the impairments in WS and DS. Although there appeared to be a length of sentence effect in the results, the data from this study did not allow for the effects of memory versus syntactic complexity to be separated fully. Sentences of longer length but with a simple, connective structure should be used as filler controls. This would allow syntax and

memory load to be investigated individually. Such sentences were removed from the present tasks after piloting because they made the test too long.

Chapter Summary

This chapter included investigations of the lexical and syntactic comprehension abilities of older children and adults with WS and DS. Using an imitation task and detailed analyses of the types of response given, it was discovered that the WS group made fewer completely unacceptable responses than the DS group and behaved more like the control groups. It appears that the individuals with WS have a more efficient language processing system than those with DS. However, they do not show the same sensitivity to grammatical structure as normal controls, as demonstrated by the subcategorisation results. Although syntactic comprehension is a problem for both groups, it does appear that the WS group exhibits a smaller discrepancy between CA and vocabulary test age than the DS group. This is further evidence that there are some aspects of language comprehension which are better than those of their counterparts with DS, but that their language skills are not fully intact as has been claimed.

The pattern of abilities found in older children and adults with WS and DS does not entirely reflect that discovered in the infant phenotype. The data from Chapter 4 indicate that both infants with WS and DS exhibit equivalent receptive vocabulary delay, whereas in this chapter it was found that the WS group performed better on a vocabulary test than the DS group. This suggests that these groups follow different developmental trajectories between infancy and the endstate.

The findings from studies of syntactic processing are less clear-cut. However, despite problems with syntax, the WS group does appear to be more sensitive to violations than the DS group and are able to remember the sentences more clearly. This slight advantage over DS for grammar appears to be present in data from toddlers with WS,

who were producing longer and more complex utterances than their DS counterparts. (Singer-Harris et al., 1997).

In the following two chapters, we turn to a different cognitive domain and investigate number understanding in infants and adults with WS and DS.

6

Specific profile: Number understanding in infants with Williams Syndrome and Down's Syndrome

Introduction

This chapter consists in its first section of an overview of the literature concerning number understanding in typically-developing infants, followed by a study of number understanding in infants with WS and DS. The literature review highlights the main number abilities present in infancy and addresses the issue of whether these abilities are number specific or whether they are the product of more general abilities. This may have a bearing on whether these abilities are impaired in the atypical groups. The second section is a report of a study examining numerical discrimination, asking whether infants with WS and DS can discriminate 2 dots from 3 dots.

Part One: Literature Review

Previous research into the early development of number understanding can be divided into studies that investigate four main areas: numerosity comparison (cardinality), magnitude judgements (ordinality), research into operations (addition/subtraction) and numerical abstraction. Although there is some overlap between these tasks, they will be discussed separately. This will allow for a survey of each of the underlying abilities that contribute to number skills in later life.

6.1 Typical Number Development

6.1.1 Numerosity comparison

This is the most investigated of all the purported numerical abilities present in infancy. Experiments test whether the infant can discriminate between stimuli which are made up of different numbers of objects, whether they be dots or pictures. This discrimination has been tested in two ways: directly, with habituation studies, and indirectly, with more complex studies which investigate infants' ability to enumerate objects and use this information to anticipate the appearance of a target. In order to make these decisions, the infant must have some way of comparing two separate sets of objects. This comparison can be made in two main ways. The first is non-numerical and relies on representations of each object in the array. In order to ascertain whether sets have the same number of members, the infant puts his representations in one-to-one correspondence with one another and uses any remainder to make the decision (Huttenlocher, Jordan & Levine, 1994; Carey, 1998; Simon, 1997; Uller, Carey, Huntley-Fenner & Klatt et al, 1999). The second method is numerical and relies on a symbolic system. The infant enumerates each set and gives the final numerosity a symbol, be it 2, --, or something even less conventional. The same is done for the other set, and then the infant compares the symbols to see if they match or if there is a discrepancy between them (e.g., Gallistel & Gelman, 1992).

Habituation studies

The first study to investigate infants' discrimination of quantity was conducted by Starkey and Cooper (1980). Infants from 4 to 7 months of age were presented with arrays of 2 dots until they reached a habituation criterion, looking less as they got bored. They were then presented with a novel number of dots (3). The infants dishabituated to the new stimulus i.e., they began to look longer again because the new numerosity was interesting to them. The arrays in the Starkey and Cooper study consisted of rows of black dots that varied in length and density but otherwise the stimuli were homogeneous. Other studies have shown that infants can discriminate numerosity even using multiple habituation stimuli which vary on several dimensions

(Strauss & Curtis, 1981; Starkey, Spelke & Gelman, 1983, 1990; Treiber & Wilcox, 1984). Infants around 10 months old can detect a change in numerosity when presented with arrays consisting of pictures of household objects even if they vary in size and configuration. The items were photographed on a 3x3 grid, so that several configurations were possible. Infants are therefore able to extract numerical information regardless of other perceptual differences. In the Strauss and Curtis study, the type of objects was varied along with size and position for some infants. They found that 10-12 month-olds could discriminate between 2 and 3, and that sometimes 3 was discriminated from 4. The ability to discriminate between 3 and 4 varied according to the gender of the infants. Females performed better when they were presented with homogeneous sets and males succeeded when heterogeneous sets were used. Strauss and Curtis (1981) suggest that this is due to the amount of attention the infants pay to the stimuli. In order to succeed with these larger numerosities the infants must be very attentive to the stimuli and it is suggested that female and male infants may be interested in different types of displays.

In sum, the studies suggest that infants can discriminate 2 from 3 and sometimes 3 from 4 but that discrimination of larger numerosities is usually beyond their capability. The ability to discriminate 2 from 3 has been found in newborns (Antell & Keating, 1983) and, from 5 months, it appears that some infants can discriminate 3 from 4 (Strauss and Curtis, 1981).

Enumeration studies

Further evidence of the ability to enumerate objects comes from studies of sequential enumeration using anticipatory saccades as a measure (Canfield & Smith, 1996; Canfield & Haith, 1991). In this paradigm the infant sees stimuli presented sequentially and, after a certain number of stimuli presented to one side, comes to expect a target on the other side. So the infant has to enumerate 1,2,3 pictures on the left before a target appears on the right. Three-month-old infants exhibit anticipatory eye movements to the position of the target after the correct number of items have been presented (Canfield & Haith, 1991). This appears to be an impressive result but

may have been due to non-numerical features of the study, e.g. the duration of the trial or the timing of appearance of the three stimuli. In order to investigate this, a further study was conducted in which inter-stimulus intervals and total trial duration were varied, meaning that numerosity alone was the predictor of the appearance of the target. Anticipatory saccades were recorded in trials where numerosity predicted the appearance of the target, but not in control trials when numerosity information was random and of no predictive value. Anticipatory behaviour was present in 3 month olds when there were 2 items presented and the infants even anticipated a target when 3 items were presented. The latter is much more demanding. To do this task, the infants must be able to "treat pictures as units whose sensory attributes are irrelevant to the prediction of the future stimulus prediction, to combine these units over time, and to use the number of items accumulated to predict the occurrence of [the stimuli]," (Canfield & Smith, 1996, p. 278). The data suggest that infants are capable of enumerating objects in sequence and, therefore, of employing some knowledge of cardinality.

In order to be successful in either the habituation studies or the enumeration study, the infant must be able to extract numerosity information. In the habituation paradigm, the infant needs to compare what he has seen with a new display and, at the very least, decide if the objects in the arrays are in one-to-one correspondence, or if the number of objects in each display differs. The infant also has to individuate each of the objects in order to perform this matching procedure. To predict the occurrence of a target in the sequential enumeration task successfully, the infant must somehow keep track of the objects that have been presented. Then, he must match this stored representation of numerosity with a criterion which tells him if a target is or is not to be expected.

6.1.2 Magnitude judgements

The section above presents evidence that supports the presence of a mechanism for cardinality in infancy. However, just because an infant can discriminate between two numerosities, e.g., 2 and 3, this does not mean that he understands the relationship between them. He does not need to know that 3 is greater than 2 nor by how much it

is greater. The studies described in this section have set out to investigate the infant's understanding of this relationship between quantities, that is, ordinality.

Evidence of ordinality understanding has been revealed in studies using 16 month olds (Curtis & Strauss, 1982, 1983) with a discrimination learning and transfer paradigm and numerosities up to 4. Infants in these studies were conditioned to respond, with a head turn, to the larger or smaller of two arrays. In the experimental trials the infants then saw either an amount they had seen previously, paired with a novel amount, or two novel amounts. In the latter case, infants must transfer their ordinal knowledge to entirely new amounts. This transfer condition also decreases the likelihood that stimulus characteristics other than numerosity are shaping responses. 12-month-old infants succeed at this task 46% of the time, whereas later in development at 16 months, infants reach criterion in 76% of cases. This suggests that there is a gradual mastery of ordinality with development.

A study conducted by Feigenson (1999) using a different methodology provides further support to these findings. Fourteen-month-olds took part in an investigation using an anticipatory looking time paradigm. The infants needed to use ordinal (less than/more than) information to anticipate the appearance of a stimulus. The findings suggest they were able to do so. Results from a habituation study (Cooper, 1984) also suggest that infants have some understanding of ordinality by the middle of their second year.

Some researchers have argued that ordinal understanding does not emerge until the child is 3 or 4 years old (e.g., Siegel, 1973). However, the task demands of the studies leading to this conclusion may have been much greater than in those mentioned in the section above. Infancy studies require only eye movements and no other action from the infant. In fact, infants do solve ordinal problems even earlier than 16 months if the items used vary on a continuous dimension. They can make size judgements such as bigger or longer, well before they realise that discrete quantities can be treated in the same way (Strauss & Cohen, 1980).

6.1.3 Numerical abstraction

There have been several studies which demonstrate that infants can use numerical information extracted from stimuli which are not purely visual. They are able to treat number as abstract and separable from its context. Wynn (1995) found that infants as young as 5 months could individuate actions and discriminate a change in the number of jumps made by a puppet. They dishabituated to a novel number of jumps even when there was continuous non-relevant movement throughout the presentation. Infants are also able to make cross modal links based on numerosity. Starkey, Spelke and Gelman (1990) demonstrated that infants between 6 and 8 months were capable of detecting correspondence between the number of drumbeats they heard and the number of dots in a visual stimulus. Both the auditory and visual stimuli were presented simultaneously and infants looked longer at the visual display which matched the number of beats they heard. It might be that these infants were making an eye movement with each beat and therefore would only appreciate the correspondence if there was a dot to match each eye gaze shift. In this way, they would pick the visual stimulus on a non-numerical physical basis. In order to try to rule out such an interpretation, Starkey and colleagues used a sequential design, in which the infant had access to only one modality at a time. This would call for a more sophisticated system of one-to-one mapping. First, infants were presented with a display of objects and then a disk appeared on the screen accompanied by either a matching or non-matching number of drumbeats. After the auditory stimulus ended, infants' looking time to the disk was measured. Six to nine month old infants looked longer at the disk when the drumbeats corresponded to the number of objects than when they did not.

However, the results described above should be treated with caution as they have not been fully replicated (Moore, Benenson, Reznick, Peterson & Kagan, 1987; Mix, Huttenlocher & Levine, 1996). In addition, some argue that cross-modal matching, such as this, is not an indication of number awareness but is, instead, an innate, domain-general matching ability (e.g., Butterworth, 1981). This might be a product of evolution, which enables the auditory and visual systems to work together to localise a stimulus. Such a process might be in operation in Starkey et al's first experiment.

6.1.4 Operations: Can infants add and subtract?

Evidence has been presented which suggests that infants are able to form numerical representations which enable them to distinguish one numerosity from another and, in some cases, to tell which numerosity is bigger or smaller. However, the data presented do not tell us whether the infants fully understand the relationship between two numerosities. They may know that 2 objects are different from 3 objects and that 3 objects are more than 2 objects but do they know how much more 3 is than 2? Karen Wynn has conducted studies that address this question, using a violation of expectation paradigm. In this type of paradigm, it is predicted that infants will look longer at an impossible or surprising outcome than at a possible one. Based on her data, Wynn (1992) argued that infants not only form numerical representations but are also able to engage in numerical reasoning by manipulating such representations. Her data suggest that 5-month-old infants are able to discriminate between incorrect and correct outcomes of the addition or removal of an item from an array. In the experiments, an infant may see one object followed by the addition of another object. Then the array is occluded and when the infant sees it again, it is either as it should be (2 items - expected outcome) or an item is missing (1 item - unexpected outcome). Infants look longer at the incorrect outcome, suggesting that they know what the correct outcome should be. This is true for the following conditions: $1+1=2/1$, $2-1=1/2$ and $1+1=2/3$. The latter condition is the most demanding because it precludes a response based only upon whether the array should have more or less items. Both the incorrect and correct outcomes contain more items. In such a case, therefore, the infant needs to know exactly how many items are present to be successful. Wynn's findings have been replicated a number of times (Simon, Hespos & Rochat, 1995; Uller, Carey, Huntley-Fenner & Klatt, 1999; Koechlin, Dehaene & Mehler, 1997). However, it appears that, as in other number studies, infants have more difficulty with this task as the resulting numerosity increases. When the outcome of the operation is 3 or greater, infants are not successful until they reach 10 months (Baillargeon, Miller & Constantino, 1994; Wynn, 1995).

Despite the attractiveness of these findings, there are alternative, perceptually based explanations for them. It may be that infants are responding to the incorrect outcome

because in addition to being arithmetically flawed, the outcome is also existentially impossible. In other words, the infants may be noticing the disappearance or sudden appearance of an object. The same type of response behaviour is seen when infants are shown physically impossible events, such as a solid object passing through another solid object (Spelke, 1991). Simon (1997) for instance, argues that no numerical knowledge is necessary to complete the addition/subtraction task. Instead, the infant is said to use four domain-general skills: the limited capacity to individuate and discriminate objects, which allows the infant to separate an array into its constituent objects; memory and comparison skills, which enable the infant to hold the objects in memory and to make a 1-to-1 mapping between the memory representation and the objects in an array; abstract encoding, which enables the infant to see two objects as an object and an object and not a doll and a teddy; and physical reasoning, or the understanding of the physical properties of objects so that the infant understands that objects do not appear or disappear magically.

6.1.5 Underlying mechanisms

The studies described above provide data that suggest that infants have a variety of numerical skills. The authors report that infants can discriminate between two small numerosities and that some can decide which is the larger of two arrays. Infants can also detect changes in numerosity when objects are added to or removed from a display. Certainly, infants display behaviour which suggests they have numerical skills. However, the mechanisms underlying this behaviour are a cause of controversy in the literature. Several researchers, such as Simon (1997) mentioned above, argue that this behaviour can be explained using non-numerical processes. A brief overview of two main approaches to this question is given below.

A non-numerical account

The infants in studies that purport to be tapping early number ability are perhaps using non-numerical means to produce what seems to be number-relevant behaviour. For example, some accounts of subitization, a mechanism that allows rapid enumeration

of small numerosities, suggest that it relies on perceptual processes. Rapid responses to small sets are made by both infants and adults (Klahr & Wallace, 1976; Trick & Pylyshyn, 1994). Mandler and Shebo's theory (1982) states that this rapid enumeration is done by a non-numerical process of pattern perception. Associations are formed between patterns and the numerosities they represent. So, for example, 2 items form a line, 3 items a triangle, and so on. It could be that infants are able to use the perceptual properties of an array to make numerosity discrimination, when two arrays of objects or shapes are presented simultaneously. This mechanism might also explain why infants are not successful when tasks involve numerosities larger than 4. This is thought to be the limit of the subitizing mechanism.

Another important non-numerical account must be considered. Several researchers have put forward models that argue that infants set up an object-file for each entity to be enumerated (Huttenlocher, Jordan & Levine, 1994; Carey, 1998; Simon, 1997; Uller et al, 1999). For example, in Wynn's study (1992) infants are claimed to form a representation of each of the dolls placed behind the screen. These object files probably begin as representations containing very little information, perhaps only the position of the object (Kahneman, Treisman & Gibbs, 1992). In order to succeed in number tasks, infants individuate objects in the array, putting each object in a new file. Then, infants compare their representations of the objects with the objects actually presented, and respond appropriately to a match or non-match. Infants are surprised if an object, which they have represented, suddenly ceases to exist. This approach reduces numerosity discrimination to physical reasoning.

Evidence for non-numerical account

The following evidence supports the non-numerical account:

- Infants have difficulties with tasks that involve numerosities over the perceptual subitizing limit - approximately 4. (e.g., Strauss & Curtis, 1981).
- When the memory load of a task is altered, performance changes. In an object file account, a higher number of objects is more difficult because more object-files must be retained in memory. If, in an addition experiment, infants are presented with objects before they are hidden by the screen, they find the task easier (Uller et

al, 1999). This is because they are able to form a stronger representation of the object/s in memory, if they have actually seen them. By a numerical account, the infant must only keep track of the final count of the set whether it be represented as 2, \$ or -- or an increment in an accumulator.

- Infants find subtraction tasks easier than addition tasks (Wynn, 1992; Baillargeon et al., 1994). This is due to the lower memory load that subtraction places on them. The infant begins with all the files he needs and has to remove some, whereas addition requires the formation of new files.
- Recent studies provide evidence that infants are using physical, non-numerical characteristics of arrays to make seemingly numerical judgements. These physical factors are contour length (Clearfield & Mix, 1999) and density (Tan & Bryant, 1996). In other words, infants are making their discriminations based on changes in a continuous variable. In the Clearfield and Mix study, infants use the total length of the perimeters of stimuli to make their discriminations and in the Tan and Bryant study infants discriminate the amount of background covered by the stimuli or the “amount of stuff”, not numerosity per se.

A numerical account

Although some researchers suggest that infants may be using perceptual cues to perform successfully in number experiments, there is evidence that supports a numerically specific account. Infants could rely on domain-general pattern matching to perform certain visual tasks, but is this possible in inter-modal perception tasks? Here, infants must compare auditory and visual information. It seems unlikely infants can rely on patterns in this case. The infants may be using another form of subitization. Some theories of subitization are numerical in nature, as opposed to that described in section 2.1.2 above. There are several theories that invoke a numerical mechanism (e.g., Dehaene & Changeux, 1993; von Glaserfeld, 1982). Wynn describes Meck and Church's (1983) animal model of enumeration, which has been revised by Gallistel and Gelman (1992) to account for number behaviour in humans. In the accumulator model, a mental mechanism produces a continuous flow of pulses at a constant rate. These pulses can enter an accumulator if a switch, which allows them

in, is closed. When items are presented the switch closes for a fixed amount of time for each item in the set. During this time, a fixed number of pulses can pass into the accumulator. The number of items can be ascertained by measuring the amount of pulses in the accumulator. The accumulator is marked with even increments, one for each item counted. In addition to the accumulator mechanism, the system has to segment the input into separate units, which can be counted, and then has to enable the input to be normalised. This means that the items to be enumerated must be stripped of perceptual qualities. This is done so that a perceptual quality of a stimulus such as the degree of brightness is not mistaken for numerosity. For example, high brightness might produce higher neural excitation and might be perceived as a larger numerosity than lower brightness which produces less excitation.

Evidence for the numerical account

The following evidence supports a numerical account:

- Infants can enumerate moving objects (van Loosbroek & Smitsman, 1990; Koechlin, Dehaene & Mehler (1997). This suggests that they might not be forming object-based static, spatial representations of the objects, but instead tagging them with some form of symbol.
- Infants can form inter-modal links between the number of objects presented and number of sounds heard, even when these are presented sequentially (Starkey et al, 1990). This might suggest that a symbolic system was being used (see discussion of this study above). It should be noted that some have argued that inter-modal perception is a fundamental sensory ability and has nothing to do with numerosity (Butterworth, 1981).
- Infants often need to attend clearly to stimuli in order to make their numerosity judgements. Infants who attended longer in Strauss and Curtis' (1981) study were successful, whereas those who paid less attention were not. This suggests that the successful infants were not using a low-level perceptual process, like that suggested by the non-numerical subitizing models.
- Infants have problems with large numerosities. These problems can be explained by the greater variance present in processing mechanisms as numerosities increase. The accumulator mechanism is a physical system in which variation occurs and, as

numbers to be enumerated increase, so does the variation (Wynn, 1995). This variation gives rise to a greater number of errors.

- However, infants can sometimes perform successfully when numerosities are high. For example, 7 month olds are able to discriminate 8 and 16 dots (Xu & Carey, 2000) and research suggests that enumeration of even larger amounts might be possible. Certainly, rats can enumerate sets up to 24 (Meck & Church, 1983). The amounts presented in these studies are too large to be handled by an object file system. They would require too much information to be held in memory, so some other system must be in operation.

6.1.6 What are infants doing and are they using number?

In my view, the behaviour exhibited by infants in previous studies can be considered as a type of numerical understanding. However, it is important to clarify what is meant by numerical. I do not believe the infants are counting, at least in the sense of the behaviour seen in children of pre-school age and beyond. The infants do not necessarily have a stably ordered set of symbols with which they tag the objects to be enumerated, for example. However, the mechanisms they use to carry out the tasks may well be the foundation of this later, numerical behaviour. For example, infants in all the studies are able to compare sets of objects using one-to-one correspondence, which is a foundation for both counting (Greeno, Riley & Gelman, 1984) and arithmetic (Kline, 1972). In order to perform one-to-one correspondence, infants must be capable of individuating the objects in some way and of stripping them of irrelevant information so that they can be treated as a set. These abilities are the building blocks for later number skills, where children must learn that any items can be counted, irrespective of their appearance. In making comparisons between sets, infants can also learn how the addition or removal of an object can have an effect on the resulting numerosity. The behaviour exhibited by infants in number studies can be thought of as precursors to later number ability, just as precursors are present in other cognitive domains. Over the course of development, infants' representations of numerosity will change to allow more complex operations, to be carried out such as

those involving fractions or zero. However, early representations could be retained for tasks involving small numerosities (e.g., subitizing in adults).

Both the numerical and non-numerical accounts of infant behaviour are supported by solid evidence. In my view, irrespective of whether infants are using one or other of these systems, they are still exhibiting the use of processes and biases which are important for numerical cognition. It may be that infants use domain-general mechanisms that are already available to them to make judgements in the early stages of number processing but that with development these evolve into more domain-specific mechanisms.

6.1.7 Literature Summary

Infants display some numerical skills, but these should be thought of as precursors to later counting and arithmetical abilities. Infants can discriminate small numerosities from one another, they can make relative size judgements, and they seem to notice a change in the numerosity of an array when an item is added or removed. However, they are not yet in possession of the full concept of number, because in order to have this they must be able to co-ordinate both ordinal and cardinal information. The number abilities described above can be attributed to infants whether or not they have physical bases, because it may be that these physical supports are present in the early stages of number, but relied on less in later life.

In the next section, a study of one of these early number skills in infants with WS and DS is presented. This will examine whether the precursors present in typically developing infants are also present in these clinical groups.

Part Two: Empirical Studies

6.2 Experiment One: Do infants with Williams Syndrome and Down's Syndrome discriminate between numerosities?

6.2.1 Introduction

There has been no published research which investigates infant number understanding in either Down's Syndrome or Williams Syndrome. However, there is a little work which examines number ability in children and adults. From this it is known that both clinical groups have difficulties with number later in life (e.g., Baroody, 1986; Bellugi, Bihrlé, Jernigan & Doherty, 1992). These difficulties are discussed in chapter 7 along with number data from my sample of older children and adults with WS and DS. Given the problems with number reported in these groups in the steady state, the following study was carried out to begin to characterise infant number ability in WS and DS. The original study consisted of two experiments examining the early stages of cardinality. The first experiment tested infants' ability to discriminate between two small numerosities (2 and 3) using a preferential looking paradigm. This task was chosen because it seemed to reflect a fundamental ability in the number domain and was performed successfully by extremely young typically-developing infants. The second experiment conducted was an investigation of inter-modal numerical abilities, using a method similar to that employed by Starkey, Spelke & Gelman (1990). In order to carry out the task it was necessary to have a numerical representation in another modality other than a visual one, which suggests a numerical rather than purely visual mechanism is present. I was interested to see if this held for infants with WS and DS as well as their typically developing counterparts. The second study was unsuccessful with these atypical infants and so is reported only in brief.

6.2.2 Method

Participants

Sixty-five infants took part in this experiment. Thirteen infants with Williams Syndrome were tested, along with 22 infants with DS, matched on both chronological age and mental age, 16 mental-age matched normal infants and 14 chronological-age matched normal infants. The mean chronological and mental ages of each group are presented in Table 6.1, and indicate a good match.

Table 6.1 Chronological and mental ages for each group

	Mean CA (months)	SD	Range	Mean MA (months)	SD	Range
WS	31	5.39	24-36	16.9	2.81	12-21
DS	30	4.90	24-36	15.6	2.43	12-20
MA	15.4	2.52	12-20	15.1	2.66	11-21
CA	30.4	5.30	24-36	30.4	5.32	25-40

Apparatus and stimuli

As in Experiment 2, Chapter 4, infants were tested using a replica of the basic Fagan apparatus (Fagan, 1970), in which two stimulus cards could be presented simultaneously. The display was illuminated by a fluorescent light positioned out of the infant's view. The centre-to-centre distance between the compartments of the display was 30.5 cm, and on all trials, the display stage was situated approximately 30.5 cm above the infant's head. In the centre of the stage was a peephole 0.625 cm in diameter, through which an observer, blind to the position of the stimuli, could see the visual fixations of the infant.

Stimuli for this experiment consisted of six white cards 17.7 x 17.7 cm. Coloured pictures of two objects were mounted on each card in different positions. The objects depicted differed on each card and included airplanes, cats, dogs, cars etc. The test cards both depicted new objects one displaying 2 items, the other 3 items. The objects on each card were of different shapes and sizes. This was done so that infants could not use merely the area of the card covered to determine which displayed the novel

numerosity. The problem of interpreting infants' behaviour as numerosity discrimination versus a more general discrimination of continuous amount has been a topic of much discussion in the literature. Therefore, appropriate controls are of great importance. For example, in a recent paper Clearfield and Mix (1999) argue that infants make their novel judgement based on the total contour length on a particular card and not on the number of items depicted. This argument has already been considered in the introductory section and will be discussed below with reference to the data from my study.

Procedure

Infants were familiarised with pairs of stimuli depicting arrays of 2 objects in different configurations and then tested with a display of 2 versus 3 objects. Each infant was tested in a special infant seat. The testing apparatus was then wheeled into position, with the display stage centred directly over the infant. At this point, the infant could no longer see the parent. The stimuli were then placed into the two compartments simultaneously by Experimenter I and, once the infant's attention was attained by talking or by shaking a rattle, the familiarisation trials began. Each infant was shown 6 familiarisation trials. After familiarisation with sets of 2, the infant was presented simultaneously with one card displaying new objects but the old numerosity (2) and one displaying new objects but a novel numerosity (3). The side on which the novel numerosity appeared was randomised and Experimenter II, who measured the cumulative looking time over each trial, was blind to the position of that card. Experimenter II held a stopwatch in each hand and timed the infant's looking to the left versus the right stimulus item by observing the corneal reflection of the stimuli in the infant's pupil. Reliability using this procedure has been shown to be high (Haaf, Brewster, de Saint Victor & Smith, 1989; O'Neill, Jacobson, & Jacobson, 1994).

A beeper was set to a fixed length for the familiarisation and test trials, and signalled when a trial was to end (10 seconds for familiarisation, 5 seconds for test). Between each trial, Experimenter I pulled back the display stage from the infant's view, recorded the data called out by Experimenter II, changed the stimuli, obtained the infant's attention, centred the infant's gaze, and finally closed the stage, exposing the

next stimuli to the infant. The roles of Experimenters I and II were randomly changed.

6.2.3 Results

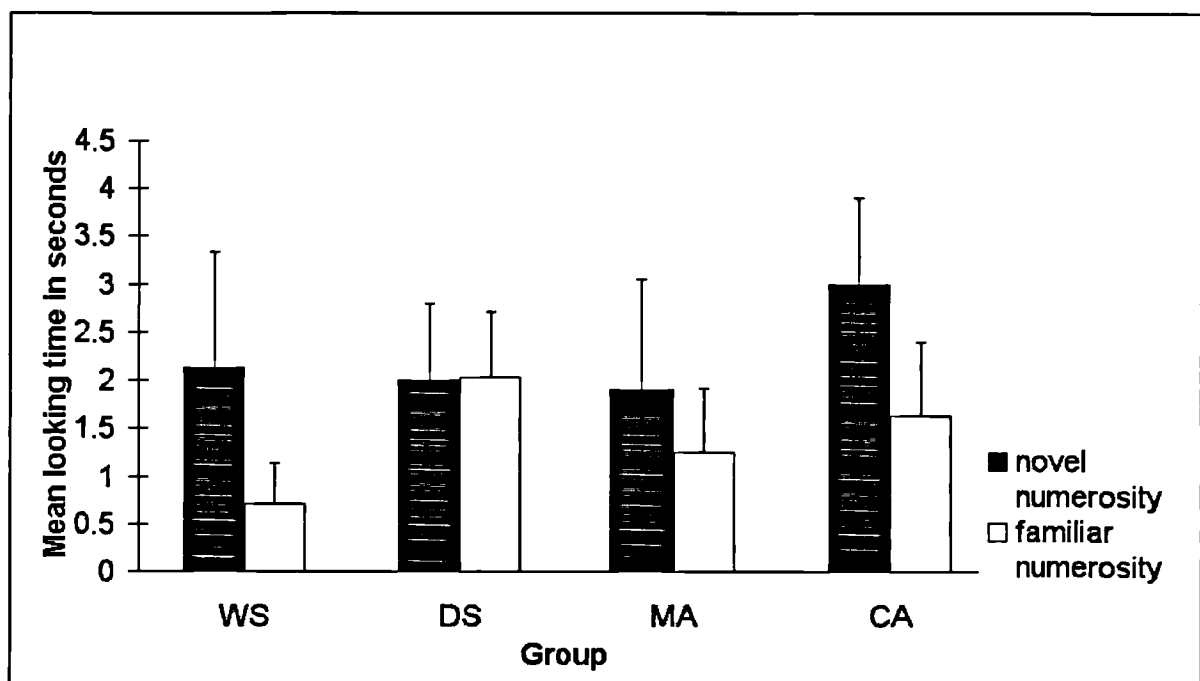
It was predicted that if infants were merely sensitive to the novelty of the objects, they would show no preferential looking because both arrays contained novel objects. However, if they had become sensitive to the constant numerosity of the displays in the familiarisation phase, then they should look significantly longer at the new numerosity.

The mean looking times to the novel (3 object pictures) and familiar (2 object pictures) numerosities were calculated for each infant. A box and whisker plot of the data from each group revealed outliers in some groups. The main body of the plot represented values between the 25th and 75th percentile. Participants whose looking time was more than 1.5 box lengths above or below these values were excluded from subsequent analyses, in accordance with the standard procedure (Kinnear & Gray, 1999). This was done to be consistent with Experiment Two, Chapter 4. This meant that data from 1 infant with WS, 2 infants with DS and from 1 MA-matched normal infant were not included in the analysis. The results were unaffected by this.

The remaining data points were entered into a repeated measures Anova with group as the between-subjects factor (WS, DS, MA-matched, CA-matched) and numerosity (novel or familiar) as the within-subjects factor. An effect of numerosity $F(1,57)=34.21, p<.001$ and of group $F(3,57)=5.94, p<.001$ was found. There was also a significant interaction of group by numerosity $F(3,57)=5.99, p<.001$. This suggests that looking time to each type of stimuli (novel or familiar) differed depending on the group. In order to investigate where the differences in performance resided, post hoc t- tests were carried out. These t-tests comparing the mean scores for novel and old numerosities for each group revealed a significant difference in looking time between numerosities for the WS and CA-matched and MA-matched groups ($t=4.2$, df 11, $p<.001$, $t=3.87$, df 13, $p<.002$ & $t=2.18$, df 14, $p<.05$, respectively). However,

there was no such difference for the DS group. The results thus show that despite their serious impairment in number in the endstate, in infancy WS participants perform normally and look like their mental-age matched and chronological-aged matched counterparts. By contrast, the DS group showed no discrimination of the difference between the novel and old numerosities. Results for the four groups of infants are presented in Figure 6.1.

Figure 6.1 Mean looking time to novel and familiar numerosities



In addition to the analysis reported above, the magnitude of difference between looking time to the novel stimuli and the familiar stimuli was examined for each group. A one way Anova, with difference (looking time to novel stimuli - looking time to familiar stimuli) as the dependent variable and group as the independent variable, was carried out. These difference scores varied significantly between groups, $F(3,60) = 5.99, p < .001$. Post hoc tests were then conducted to compare the magnitude of these differences for each group. There was a significant difference in the magnitude between the WS and DS groups, Tukey HSD ($p < .05$). The WS group difference was larger than the DS group difference. The magnitude of difference for the CA-matched group and DS group were also significantly different, Tukey HSD

($p < .05$), with the CA group exhibiting a larger difference than the DS group. There were no other differences.

6.2.4 Discussion

The present study provided the first data ever to characterise the performance of infants with DS and WS using a numerosity discrimination task. Typically developing infants, whether mental age or chronological age-matched, looked significantly longer at the arrays of a novel numerosity (3) after familiarisation with arrays of 2. The WS infants performed like their typically developing counterparts, whereas the DS infants did not. This is particularly interesting, given the impaired pattern of abilities reported for these atypical groups in older childhood and adulthood.

There have been reports which suggest that adults with WS have particular difficulty with number, in the face of their relatively spared language (Bellugi et al, 1992). There is also experimental evidence to show that they are unable to conserve number (Bellugi, Marks, Bihrlé & Sabo, 1988). This ability to conserve is present in typically developing 6-7 year olds. Despite these problems, evidence from the present study suggests that one of the fundamental underpinnings of number understanding is present and working in WS infants. This is the ability to discriminate small numerosities.

Although the present study provides only one test of infant number ability, it is a test which has been replicated a number of times in a variety of forms (e.g., Strauss & Curtis, 1981; Starkey, Spelke & Gleman, 1990). On the basis of the data, it seems again that the pattern of cross-syndrome performance in the steady state of late childhood and adulthood is not reflected in the pattern of abilities of the different atypically developing infant groups. The WS group is able to extract numerical information from the displays, despite later problems with numerical tasks, while the DS group does not demonstrate this ability. The infants with DS may have impairment in a basic ability which underlies later number ability. However, there are

other possible reasons for the lack of difference in their looking times that must be ruled out first. Failure to habituate to the familiar numerosity could have caused no difference in looking time at test. In this study, this is unlikely to be an explanation because looking time in both the WS and DS groups decreased by a similar amount across the familiarisation trials. (The mean differences between looking times to the first pair of stimuli and the last pair of stimuli were calculated for each individual in the DS and WS groups. An Anova indicated no significant difference in the decrease of looking times across groups, $F(23,36) = 1.16$, $p > .40$. The mean difference in looking time for WS was 1.49 secs, SD 3.13; and for DS was 1.39 secs, SD 3.38). It is also possible that attentional differences may have led to the even pattern of looks to both familiar and novel stimuli in the DS group. Work with typical infants has shown that differences in attention to stimuli may lead to different results in numerosity experiments (Strauss & Curtis, 1981). However, infants with DS showed preferential looking behaviour in the language study and in a pilot study involving interesting patterns versus a grey patch, which required attention to both stimuli. In the present study, it is likely that these problems can be ruled out. It seems, therefore, that one aspect of the precursors of number, sensitivity to changes in numerosity is present in infants with WS but not in those with DS.

This means that the developmental trajectory for number understanding in these two clinical groups differs. In DS, problems with number appear to reside in its foundations and are apparent very early in development. By contrast, in WS it seems that at least one of the foundations of number understanding is functioning normally. Either the root of later problems in this group occurs further along the developmental pathway or perhaps another of the fundamental building blocks for number might not function appropriately. As is seen in language development, it may be that the DS infants are delayed in their acquisition of these precursors of number, and it is the delay that leads to their difficulties. By contrast, in the WS case the infants may have parts of the foundations of number but may acquire subsequent number skills by a different route. Further research following the same infants longitudinally over time should enable us to discover when the number problems encountered by individuals begin and in which tasks they are encountered.

6.2.5 Future directions and concerns with the present study

The study presented above was the first of its kind. A great deal more research must be carried out to characterise further the number abilities of infants with WS and DS. Infants should be tested using all the paradigms mentioned in the introduction above. These should include more numerosity comparison studies, tests of ordinal ability and those which involve “addition” or “subtraction”. In addition, refinements could be made to the test used in the present study. First, in the present study all infants were familiarised with 2-item arrays, making the 3-item array the novel stimulus. In a replication, half the infants should be familiarised with 3 and tested with 2 v 3 and half should be familiarised with 2 and tested with 2 v 3. In the past, concerns have been raised that infants look longer at the 3 objects, not because of novelty but because they are just more numerous. I believe this not to be the case, because several studies have shown that infants do not show a baseline preference for 3 objects over 2 objects (e.g., Wynn, 1992), but a design including both conditions would be a safeguard against this potential problem. Second, the stimuli used in the study could be further refined, in the light of the ongoing debate over numerosity versus the amount of space filled by the stimuli. In the present study, attempts were made to have two items cover the same area as three, but this was not done with absolute precision. A further study using computerised presentation could balance the stimuli more accurately.

6.3 Experiment Two: Inter-modal numerosity comparison

This experiment examined the ability of infants with WS and DS to extract numerical information from both auditory and visual stimuli and to compare these numerosities. Infants were tested using a preferential looking paradigm. As done in Experiment 1 the Fagan apparatus was used to present stimuli, comprising of arrays of either 2 or 3 objects. There were 6 test trials in which an array of 2 items was presented together with an array of 3 items. The side of presentation was varied, with each stimulus presented an equal number of times on the left and the right. Once the infant had been presented with the visual stimuli, the auditory stimulus began. This consisted of either

2 or 3 beats played on a glockenspiel. Again, 3 trials consisted of 2 beats and 3 trials consisted of 3 beats. The trials were randomised, with the last 3 trials as a mirror image of the first three. Each trial lasted 5 seconds and looking time to each stimulus was measured, using the same procedure as in Experiment 1.

It was predicted that infants would show a pattern of preferential looking and, on the basis of Starkey et al (1990), that they would look longer at the display with the same number of objects as the beats they heard. Usually in cross-modal studies infants prefer familiarity rather than novelty. Of course, it was also possible that they might look longer at the novel display, i.e., at the items which did not correspond to the beats, as reported by Moore et al. (1987). The preliminary data collected for this study were, alas, inconclusive. The task did not appear to be working with my sample of DS and WS infants nor, more importantly, with the typically developing controls. Their looks to stimuli matching and non-matching were erratic. It was therefore decided to discontinue testing, given the inconclusive data which have been collected from typically developing infants in the existing literature.

Future research could address this task, using two different paradigms: one in which auditory and visual stimuli are presented simultaneously, and another in which stimuli are presented sequentially, beginning with a display of objects followed by a disk on the screen accompanied by either a matching or non-matching number of drumbeats. These two paradigms could help to ascertain whether any conclusive data could be gathered from the DS and WS groups.

Chapter Summary

The first study of number understanding with WS and DS infants was presented in this chapter. It was found that the WS infants were able to extract numerosity information from familiarisation stimuli and use this to discriminate between two new displays depicting one novel numerosity (3) and one familiar numerosity (2). This was not the case for the DS group who appeared not to demonstrate this ability. The results of the study suggest that, despite reported problems with number in later life in

both WS and DS, the infant profiles of these groups differ. WS infants appear to have at least one of the precursors to number in place in infancy, whereas this precursor is not demonstrated by the DS infants. Again, this highlights the importance of investigating infant precursors to later abilities. If adult profiles on number tasks which tap the mature representation of number were used to infer infant performance it might be concluded that both groups would have number problems in infancy. Instead, the present study suggests that number processes follow different trajectories in each group.

The following chapter consists of an examination of number abilities in the endstate.

Specific profile: Number understanding in older children and adults with Williams Syndrome and Down's Syndrome

Introduction

This chapter is an examination of numerical competence in older individuals with Williams Syndrome and Down's Syndrome. The development of number ability is of particular interest because it is often reported as causing problems for both of these clinical groups. In Chapter 6, infant competence in the number domain was explored. A preferential looking experiment revealed that infants with WS could discriminate between a familiar and novel numerosity, whereas the infants in the DS group did not demonstrate this ability. We now investigate whether this cross-syndrome difference holds in the phenotypic outcome.

The chapter is in two parts. The first provides an overview of some of the number literature in normal and atypical development. It describes the counting principles employed by typically developing pre-schoolers and is followed by an overview of some important elements of arithmetic development. Finally, the performance of typically developing individuals on number comparison tasks will be considered. These areas of number competence were chosen because they have also been investigated in atypical populations, and are therefore important when making comparisons across groups. In addition, a description of the symbolic distance effect is provided because this task forms the main body of the empirical work reported in

Part Two. The overview continues with a consideration of the little published research there is on WS number, and is followed by an overview of the existing data from studies of DS.

In Part Two, 4 studies are reported. These involve older children and adults with WS and DS, along with school children with chronological ages similar to the mental ages of the WS and DS groups (MA-matched group), and another group of a similar chronological age (CA-matched group). The first 3 studies form an investigation into the representation of numerosity in each group. This is thought to be an indicator of an understanding of the meaning of numbers. In a number of respects, it is similar to the task used in the chapter on infant number understanding, involving the processing of two numerosities. The performance of individuals with WS and DS on this task should give an indication of whether the important principles underlying number understanding are in place. The second study is an assessment of number understanding and basic arithmetic, using a battery of number tasks usually employed with neuropsychological patients. This allows for a comparison across groups on a variety of number tasks.

Part One: Literature Review

7.1 Typical number development: counting

This section will provide a brief description of the main principles used by typically developing individuals to constrain their counting behaviour. Although the research has been carried out with pre-schoolers, it is given consideration here because tasks tapping counting principles are among the only ones to have been used with individuals with DS and WS.

In order to class a child's number ability as true counting, it is necessary to define the exact nature of the counting process. Gelman and Gallistel (1978) suggest that certain counting principles must be adhered to in order for behaviours, which look like counting on the surface, to be instances of true counting. As happens in lexical development, such principles enable the child to constrain their processing of input from the environment to that which is appropriate to the number domain, and to distinguish this from non-numerical input. There is much debate as to whether innate counting principles shape later counting (Gelman & Gallistel, 1978) or whether repeated exposure to counting by rote allows children to subsequently extract the principles (Fuson & Hall, 1983; Fuson, 1988). However, of interest here is the nature of the principles.

The following principles are those which begin to guide typical counting behaviour in the pre-school years. They are most often assessed using a novel counting task, which will be considered below.

One-to-one correspondence or unique tag principle: Each item to be counted is given a unique tag and is counted only once. So, for example, if a pair of shoes is to be counted, although the array is made up of /shoe/ /shoe/, it would be counted using two different labels "one, two".

Stable ordering principle: Tags are always applied to items in the same, stable order. This does not have to be the conventional order. The count 1,2,5,3,8 is acceptable provided it is used consistently by a child.

Item indifference: Any item can be counted, for example, sounds or separate pieces of a broken object (Shipley & Shepperson, 1990).

Order irrelevance: Items can be counted in any order, as long as all items are counted once. The count can go from right to left, left to right, or start in the middle of an array (Gelman & Cohen, 1988).

Cardinal principle: The last tag of a count denotes the cardinal value of the set. However, this must involve more than the mere rote repetition of the last tag. Competence with cardinality has to be demonstrated even when the cardinal value is not provided in the experiment, for immediate repetition. In other words, the child must be able to work out the cardinality and not just repeat the last tag of a count that was provided for them. E.g., if they are given the count 1,2,3,4 the response “4” does not constitute an understanding of cardinality. The latter involves forming a link between the counting process and a conceptualisation of numerosity, the precursors of which are seen in infancy. Wynn (1990) demonstrated that, when asked for a specific number of objects from a large pile, by 3.5 years children were able to give the correct number of items to an experimenter. However, a younger group (2.7 years) merely grabbed a random number from the pile, despite the fact they knew how to count. The latter group had not yet realised the full meaning of the link between counting and cardinality.

The use of counting principles can be assessed using the novel counting paradigm in which normal rote counting strategies cannot be adopted. In the constrained counting task (Gelman & Gallistel 1978; Gelman, Meck & Merkin, 1986; Gelman & Cohen, 1988), children are asked to count an array of five objects making sure that a particular object (pointed out to the child) is given a pre-specified tag. So, for example, in a line of 5 objects the child is told the third object must be termed “the

two". To succeed, the child must come up with a novel solution to the counting problem but still adhere to the counting principles. Four and 5 year olds succeed on this task. They therefore show an implicit knowledge, at least, of the principles that constrain counting. They are able to improve their own performance with practice and can notice and correct their own errors as well as benefiting from external hints. This benefit could only come from an existing understanding of the principles that would allow for the use of strategies to surmount problems. An even more sophisticated instance of this understanding is seen in the importance placed on the adherence to each principle when coming up with a solution. Some children who made errors on the task nevertheless showed quite sophisticated understanding of the principles. They knew that the one-to-one principle, giving only one tag to each item, was of fundamental importance in counting but that labelling was more flexible. These children would alter the sequence of labels, e.g., 1,3,2,4,5,7 but would not count an object twice nor miss one out, knowing that omitting an object or doubling up would also have consequences for the resulting cardinal value. They knew that if they did this they would give the wrong total number of objects and would have violated the one-to-one principle. Such understanding of counting principles is also shown in tasks involving the detection of errors in another's counting (Gelman & Meck, 1983; Gelman, Meck & Merkin, 1986). However, it is important to treat these results with caution, given the problems with interpretation of cardinality understanding mentioned above. In the sections on WS and DS number, we will return to counting behaviour to examine whether the older clinical groups behave like typically developing pre-schoolers. The following section will consider some aspects of arithmetic which have relevance to the performance of these clinical groups.

7.2 Typical number development: arithmetic

The ability to carry out simple arithmetic is an extremely important skill for everyday life, for example, when dealing with money. This section will provide an overview of the development of arithmetic skills in typically developing children and will make suggestions as to the cognitive processes that underpin them. We will see that this

literature challenges some of the interpretations of infant capacities discussed in chapter 6.

There are several non-numerical abilities that are important for the development of number: symbolic representation, spatial representation, language, memory and processing speed. As we shall see, if any of these are impaired, even alongside good numerosity capacities, the development of number will be impaired. It is also possible that different syndromes will display similar number impairments but that these are due to different impairments in non-numerical abilities, such as language or memory.

7.2.1 Non-verbal reasoning and arithmetic

The chapter on infant number discussed the possible presence of addition and subtraction processes in pre-verbal infants and whether the skills were specific to number or domain general. Huttenlocher, Jordan & Levine (1994) argue that even in older toddlers, mental representations based upon domain-general abilities are used to solve non-verbal addition problems. They found that the ability to form mental representations, such as creating a spatial model of a room (DeLoache, 1991) or substituting objects to stand for other objects in pretend play (e.g. McCune-Nicholich, 1981) emerges in toddlers at about 2½ years old, and that it coincides with the ability to solve non-verbal arithmetic tasks with small quantities (up to about 5). This is why they claim that the ability is domain general. In their study, children were asked to perform operations on a set of hidden objects to make them match the experimenter's objects numerically. A child would be shown two objects which were then covered. Following this, two further objects were added. The child had to choose the appropriate number of items from their own set to match the result of the experimenter's sum. Two and half-year-olds could do this successfully. Huttenlocher et al. argue that where infants may be using a system of approximation to guide their numerical behaviour, their findings suggest that toddlers are beginning to use a symbolic system to represent number.

7.2.2 Language and arithmetic

As children develop their use of symbols, these allow them to read off the number line more accurately because they give a specific label to numerosity. In turn, this will lead to greater success on number tasks. This could be the first step on the road to verbal arithmetic, with children moving from mental symbols which represent objects, to the appropriate verbal or visual number label. Levine et al. (1992) illustrate this progression, finding that 4 year olds can solve number-related non-verbal problems, but not number-related story problems which are verbal. However, 5 year olds solve both types of problem. Once children begin to solve verbal problems, or use verbal means to solve those presented in the Arabic numeral form, language skills begin to have an influence on mathematical achievement.

Language impairments can cause problems in number even in children of normal intelligence. For example, the link between language and number is evident in children with specific language impairment, who often have concurrent difficulties with mathematics. Number conservation and classification are impaired in such children (Camarata, Newhoff & Rugg, 1985; Kamhin, 1991). It is likely that part of their difficulties stems from problems with verbal short-term memory (Fazio, 1996).

7.2.3 Memory and arithmetic

As children's arithmetical competence develops, so does their strategy use (e.g., Siegler 1986, 1991). Children begin solving addition problems by counting all the items to be summed (Baroody & Ginsburg, 1986). They then learn to count on from the first number (e.g., Fuson, 1988). After practice, the solutions to problems are stored in memory and retrieved when needed (Resnick, 1983). It is at this stage of the development of number that memory ability becomes particularly important.

Memory impairments can cause problems in two ways. First, they can lead to problems with retrieval of number facts (Fazio, 1996) and second, they may disrupt the calculation procedure itself. In order to perform a mental calculation, a child must hold 2 quantities in memory, retrieve the appropriate operation and then carry out the

calculation (Logie, Gilhooly & Wynn, 1994). It is known that adolescents who are very good at maths have superior short-term memory and do particularly well on tasks involving spatial locations and digits (Dark & Benbow, 1991). The need for both long-term memory (for fact retrieval) and short-term memory (for the encoding of these facts and use in calculation) might have implications for the performance of individuals with WS and DS, as it is known that these groups have memory impairments. The results concerning memory for spatial location may also have particular relevance to the performance of the WS group. Although individuals with WS do seem to be able to move around their environments with little problem (Mervis, Morris, Bertrand & Robinson, 1999), their other spatial deficits might have an impact on their representational skills for number.

7.2.4 Speed of processing and arithmetic

Another important contributor to performance on arithmetic tasks may be processing speed. Bull and Johnston (1997) found that when a group of 7 year olds was divided into two sub-groups using standardised tests: 1) those good at maths and 2) those poor at maths, processing speed and item identification were predictors of ability. In their study, processing speed was measured using both a perceptual motor task and a visual number-matching task. Bull and Johnston suggest that slow processing speed could lead to lower efficiency of short-term memory which, in turn, means that number facts are poorly encoded in long-term memory. This weak encoding might lead to less automaticity in number fact retrieval. The finding that children who are poor at maths are slow to recognise Arabic numerals, also points to a lack of automaticity (Hitch & McAuley, 1991).

Could it be that the number difficulties exhibited by individuals with WS and DS are due to a domain-general information processing impairment and not to the number domain per se? Or are there particular areas of number, which show greater impairment than others? This question remains open. Dehaene, Spelke, Pinel, Stenescu and Tsvikin (1999) suggest that different aspects of number manipulation follow distinct developmental trajectories, so it is possible that some but not all of

these are affected in WS and/or DS. They propose one pathway for numerosity encoding, analogous to a mental number line and used for tasks like approximate calculation, and another for symbolic manipulation and exact calculation. Interestingly, they suggest that it is the dorsal pathway, with its visuo-spatial processes, which may be used for the approximate calculation and numerosity encoding processes. Some research has suggested that this pathway may be impaired in individuals with learning difficulties, particularly those with WS (Atkinson, King, Braddick, Nokes, Anker & Braddick, 1997).

Now that some aspects of arithmetic have been considered, an overview of the literature concerning number comparison will be presented.

7.3 Typical development: number comparison

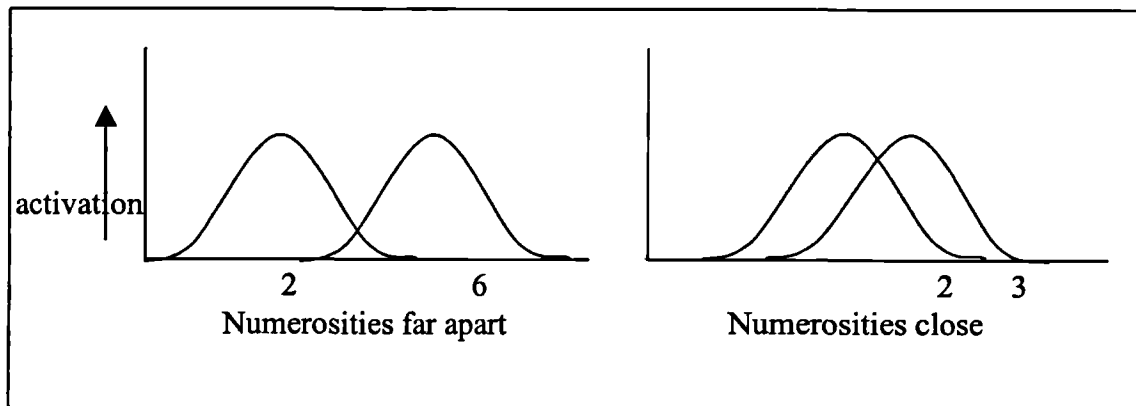
Number relevant information is presented in a variety of ways. One must be able to deal with: (1) the visual Arabic system, e.g., 7 or 56; (2) the verbal number system, e.g., /seven/ or /fifty six/; and (3) an analogue magnitude code, in which numerosity is represented on some kind of mental number line. The last of these is likely to be available early in development, as discussed in Chapter 6. It is a pre-verbal system which is available to rats and pigeons as well as human infants (Washburn & Rumbaugh, 1991).

The presence of an analogue mechanism with which magnitude is represented can be demonstrated using numerosity comparison tasks. These tasks tap the mapping between magnitude codes and their other labels. To use numbers effectively, one must be able to partition the magnitude representation in order to assign number labels appropriately. One must also have a concept of the magnitude which a verbal or Arabic label represents. In a numerosity comparison task, the participant is presented with two numerosities or Arabic numerals and is asked to press a key that corresponds to the larger. Reaction times are measured, to investigate the effects of different distances between numerosities.

This task has given rise to a very robust effect, known as the symbolic distance effect (Moyer & Landauer, 1973). Participants take longer to discriminate between numerosities which are close together, e.g. 3 and 4, than those which are far apart, 2 and 7. It turns out that the time taken to compare two numbers is inversely proportional to the numerical distance between them (Moyer & Landauer, 1967).

This effect is also found in psychophysics with judgements about dimensions, such as volume or length, which suggests that participants are reducing symbols to some kind of magnitude representation (Moyer & Landauer, 1973; Buckley & Gillman, 1974). The effect has also been found using Arabic numerals, dots and verbal number labels as stimuli. The cause of this effect is likely to be the variability in the magnitude representation. If the mental number line is rather fuzzy (Dehaene & Cohen, 1994), then it will be easier to distinguish the larger of two numbers if they are far apart and have little possibility of overlapping activation (Figure 7.1 below).

Figure 7.1 Variability in magnitude representation



Some researchers suggest that all number codes are converted into a magnitude code in order to process them further (Dehaene, 1992; McCloskey, 1992). Dehaene argues that the magnitude code holds the semantic information for a number symbol and will be called upon when number symbols are not useful, for example in approximation and estimation. For example, Dehaene and Cohen's (1991) patient NAU was unable to perform calculations using symbols but could make magnitude comparisons and call upon these to do arithmetic.

The distance effect is very robust. It is found in the performance of young children from at least 6 years of age (Sekuler & Mierkiewicz, 1977; Duncan & McFarland, 1980) and, as mentioned above, is based on a pre-verbal code. Research with adults has added support to this, by showing that the distance effect is present in studies which use two languages with very different number symbols, e.g. English and Iranian (Dehaene, Bossini & Giraux, 1993)

In sum, it appears that the symbolic distance effect is a behavioural indicator of a fundamental part of the number system, i.e., the understanding of the quantity that numerical symbols represent. It is therefore important to investigate whether this effect is present for individuals with WS and DS. An absence of the effect might suggest that there is a fundamental impairment in number understanding.

Now that aspects of number ability in the normal population that have relevance to the data from clinical populations have been considered, an overview of the data from number studies on WS and DS will be given.

7.4 Number ability in Williams Syndrome

There are many anecdotal reports concerning the numerical problems that people with WS encounter. Many parents are concerned about their children's difficulties with basic arithmetic and counting, especially when it interferes with basic activities like shopping and using public transport. However, very few systematic investigations of numerical competence in WS have been carried out. The few data available from studies that have considered number ability in WS are described in the two sections below.

7.4.1 Williams Syndrome: counting and number concepts

There are only two studies which include an investigation of counting behaviour in WS. Karmiloff-Smith (1992b) carried out a small study of 4 adolescents with WS.

She found that, despite being able to count by rote, they could not successfully complete a relatively simple counting task, which involved counting a circular array of 9 objects. Their difficulties could not be attributed to memory impairment, because their performance remained poor even when a marker was provided to indicate where they started their count. The performance of adolescents in the study was extremely poor, given that typically developing 5 year olds should be able to perform this task (Gold, 1989).

The second investigation of counting in WS was an undergraduate project by Hughes (1995). He investigated Gelman & Gallistel's (1978) counting principles in WS. Hughes' study was more informative than many, given that it used both normal pre-schoolers, divided into two subgroups: 4 year olds and 5 year olds, and 11 WS subjects aged 8-33 years. The findings could therefore highlight differences in the use of counting principles between the two main groups. The subjects underwent a series of tasks examining the counting principles postulated to be building blocks for later numerical cognition and were asked to count both objects and sounds. Sounds were used in order to probe the subjects' ability to generalise these counting principles, to circumvent their spatial difficulties, and also to prevent participants recounting the stimuli. It was found that individuals with WS abided by all the counting principles used by normally developing 4 and 5 year olds in tasks using both objects and sounds as the counting stimuli. Participants with WS could also detect errors in someone else's counting which violated one of the three counting principles: stable order, one-to-one correspondence or cardinality. This suggested they were sensitive to these principles and indeed performed more accurately when performing counts themselves than the typically developing pre-schoolers. All groups found objects easier to count than sounds, but the difference in performance was considerably less marked in the WS group. This is interesting because it casts doubt upon the hypothesis that poor counting in WS may be due solely to visuo-spatial difficulties (Bellugi et al., 1988) because the WS still find object counting easier than sound counting, which was supposed to ameliorate any spatial problems. However, the fact that object counting was not greatly better than sound counting does suggest that visuospatial difficulties might make some contribution to WS number problems.

The seemingly intact ability to count sounds is interesting for a completely different reason. Sometimes the presence of ability is as informative as impairment and may point to a difference in the process of development. This could be the case with counting sounds. It appears that the typically developing group has an advantage with objects, which are the entities most usually counted. However, this is less marked in the WS group. The small change in the constraints used by this group may not have large effects in a simple task. However, a small difference in the fundamental constraints used may lead to more serious problems when the demands on the number processing system increase.

In the second of Hughes' experiments, the participant's own counting ability was tested both for objects (which could be recounted) and sounds (which could not be). Three amounts were counted by each child for each modality. It was found that individuals with WS performed almost at ceiling when counting objects and at 83.33% correct in the sound trials. They also counted significantly more accurately than the other two groups of younger typically developing children. There were unfortunately no mental age matched controls.

In order to give a more stringent test of the understanding of cardinality, the subjects also undertook a "Give a number task" in which they were asked to hand the experimenter a certain number of objects from a large pile (Wynn, 1990) or, in the sound condition, to produce the appropriate number of tones. This task should be a good indicator of understanding of the cardinality principle as the subject has to be aware of the final amount of objects they have, to know that counting is relevant to the successful solution, and not just something one does when asked "How many?" It was found that the WS group was the most accurate counters and that both they and the 5 year old normal group understood the cardinality principle, whereas 4 year old children seemed to have difficulty with the task.

However, when considering the results it should be noted that the WS subjects were of higher MA (measured using Ravens Progressive Matrices) than either control

group. In order to investigate the development of counting principles more thoroughly, a more closely MA-matched control group is essential. The Hughes study grouped together both WS children and adult subjects which again makes it difficult to examine the developmental pathway itself. That said, the study does give an early indication that in number, unlike in lexical development, individuals with WS seem to follow all those principles used by normally developing children. This finding raises the issue of the origin of the difficulty seen in later numerical skills in individuals with WS. The use of more demanding number tasks differentiates between individuals with Williams syndrome and normal subjects. These tasks include Piagetian number conservation and seem unlikely to arise from a lack of use or understanding of fundamental counting principles. The difficulties encountered by people with WS on these tasks must be due to another aspect of number understanding or perhaps even another aspect of cognition.

7.4.2 Williams Syndrome: number conservation

On the basis of number conservation data, Bellugi, Bihrlé and Sabo (1988) argued that individuals with WS have severe number impairments. However, the research that they report has a very small number of participants and needs to be expanded. Nevertheless, they found that individuals with WS have difficulty with Piagetian number conservation tasks. Typically developing children of 6 or 7 years are able to perform these tasks successfully, understanding that a change in a physical attribute of an array does not affect its numerosity. However, individuals with WS cannot. Incidentally, one of our brightest participants with WS did not realise that 2 rows of paper clips contained the same number of items when one of the rows was spread out further than the other. He was convinced that the widely spaced row contained more clips. Bellugi and her colleagues suggest that individuals with WS may fail this task because they are unable to ignore irrelevant perceptual details when making their decision, and are fooled into picking the more widely spaced row. Since it has been demonstrated that individuals with WS have problems with visuo-spatial constructive tasks (Pani, Mervis & Robinson, 1999), it could well be that the perceptual irrelevance hypothesis is correct. Pani et al. argue that individuals with WS may have

difficulty switching between different levels of spatial organisation, for example from global to local. This could be the problem in conservation tasks where the participant sees a global change (a longer line) but does not then switch to see the number of local elements (paper clips) remains the same.

7.4.3 Williams Syndrome: arithmetic

In addition to the research examining basic number processes, such as counting, there has been one study which included measures of arithmetical ability. Udwin, Davies and Howlin (1996) measured this ability in a longitudinal study of cognitive skills. They found that at the mean chronological age of 12 years, all the participants had a test age of 99.6 months (8;3 years) on the arithmetic subscale of the WISC R, and a test age of 97.2 months (8;1 years) at a mean age of 21 years on the WISC III. They suggest that the drop in arithmetic performance was unlikely to be the result of developmental change, but due to the slightly different tests used on each occasion. The WISC III, which was used for the second round of testing, demands a higher level of performance for a similar test age than does the WISC R, which was used for the first round. However, despite the scoring problems, these results indicate that performance on arithmetic scales remains at a plateau, at a level about 7 years below chronological age. This difference is not due to general cognitive impairment, because it is much greater than the lag between CA and language and CA and overall MA. Udwin et al.'s study reveals true impairment in numerical ability but does not examine individual numerical abilities in their own right, nor does it investigate the particular bases of the impairment. This type of investigation has yet to be carried out.

From this overview, it is evident that even in the face of intact counting skills, individuals with WS do have problems with several aspects of number, such as conservation and arithmetic. In the empirical section of this chapter, an investigation of fundamental number understanding and basic arithmetic will be described, but first we turn to number abilities in Down's Syndrome.

7.5 Number ability in Down's Syndrome

More research has been conducted into numerical competence in DS than WS. The following sections will concentrate on what is known about counting and arithmetic ability in DS.

7.5.1 Down's Syndrome: counting

There have been a number of studies examining the counting processes of children with DS. Early studies suggested that individuals with DS could count by rote, but showed no understanding, and that they exhibited no understanding of arithmetic concepts (Cornwell, 1974). By contrast, more recent research has uncovered greater competence in the DS population. Gelman & Cohen (1998) examined the use of counting principles in 10 children with DS (MAs 3;6 years - 6;8 years) and in a group of 10 pre-schoolers (4-5 years). They found that the children with DS did not perform as well as the control group in a novel counting task. This task requires the child to have an explicit understanding of counting principles and in particular taps that of order irrelevance. The child must count an array in an unconventional order and understand that no matter how it is counted, the number of items remains the same. Most of the children with DS in the Gelman and Cohen study were rote counters, but two were classified as excellent, and could complete the novel counting task by using flexible counting strategies.

Unfortunately, the matching procedure in the above study was rather imprecise, but this was improved upon by Caycho, Gunn & Siegal (1991). They matched a group of children with DS (mean CA 9;7) with a group pre-schoolers on the basis of a test of receptive vocabulary (the Peabody Picture Vocabulary Test, PPVT). Both groups were at the same developmental level, i.e., at a test age of 4;7 years. Caycho and colleagues found that developmental level and the presence or absence of DS predicted performance on tasks involving Gelman & Galistell's counting principles. The majority of the children with DS could employ the 1-to-1 correspondence, stable order, and abstraction principles. They could also answer the cardinal type question "How many?" Those children who successfully completed the novel counting task

had a vocabulary test age of 4 years or more. This fits with Gelman & Galistell's proposal that explicit understanding of counting principles emerges at 3 years and is fully developed by 5 years.

7.5.2 Down's Syndrome: general numerical competence and arithmetic

Studies of counting have suggested that developmental level and not Down's Syndrome per se, is a good indicator of success. There are mixed results regarding the contribution of mental age level to arithmetical ability. Sloper, Cunningham, Turner and Knussen (1990) conducted a study of 117 children aged 6-14 using a teacher questionnaire about mathematical ability. Items on the questionnaire covered a wide range of skills, from those which assessed numerosity discrimination (can pick the larger of two arrays) to arithmetic (simple division sums). Mental Age correlated .73 with performance as assessed by the teacher in this study. As would be expected the type of education the child was given had an effect on performance, with those in mainstream schools doing better than those in special schools. This is supported by findings from Casey et al. (1988). However, in contrast to the findings of Sloper and colleagues, a study of a group of children with mixed aetiology mental retardation reported that some of the children with low IQs (33-49) performed better on a number battery, based on what is expected of an average 6 year old in the USA, whereas others with higher IQs (51-80) were amongst the poorest performers (Baroody, 1986). It should be noted that Baroody's sample was a mixed aetiology group, but it does suggest that IQ might not be the best indicator of the numerical capabilities or potential of an individual, especially given the importance of the educational background of the child. Many children with DS are given very little numerical instruction (Bird and Buckley, 1994), and this may be one contributor to their number difficulties.

In a recent more detailed study, Nye, Clibbens and Bird (1995) used the instruments from both the Sloper et al. (1990) and Baroody (1986) studies. They found that the two measures correlated highly when used with 16 children with DS aged 7 years -

12;6 years, so it is likely that the measures were tapping similar numerical abilities. The tests also correlated with standardised measures of numerical competence from the BAS and the Kaufman Assessment Battery for Children. Performance of children with DS on number tasks was significantly related to their grammatical comprehension, as measured by the TROG (Bishop, 1983), but did not correlate significantly with receptive vocabulary. This result is likely to be due to the type of numerical skills investigated in each study. Different numerical skills draw upon language to different degrees. Dehaene (1992) suggests that while numerosity comparison has no verbal element, arithmetic and counting draw upon language capacities. Within the number domain, it may be that different tasks call upon different verbal skills.

An important finding of this study is that performance varies a great deal across individuals. Older children with DS do not necessarily perform better than younger ones. Instead, performance is likely to be related to the input they have received. It is certainly the case that intervention studies have an effect on numerical skills. For example, Irwin (1991) found that if a child with DS could count, then he could be taught how to use the counting-on strategy for addition problems. Baroody (1988) also showed that children with mental retardation could invent their own addition strategies if given a great deal of practice with addition. The results relating to variability of performance and the type of input provided are of importance when considering the performance of all atypical groups. They have relevance to the empirical work reported in a later section of this chapter.

In summary, then, it appears that children with DS are impaired in the number domain, and in some cases this impairment can be linked to the developmental level of the child. Children with DS can master the principles of counting but may have greater difficulty with more complicated arithmetic.

7.6 Literature summary

Both the individuals with Williams Syndrome and those with Down's Syndrome can use the general counting principles proposed by Gelman and Gallistel (1978). For the DS group it appears that this ability is linked in some way to developmental level and that language is important in number tasks. The existing data do not permit a similar conclusion to be drawn for individuals with WS, but the studies reported in part two of this chapter may shed light on the link between language and number in this group. Despite having the ability to count, at least to some degree, the previous research indicates that the WS and DS groups both have difficulty with number. The following section will further investigate number ability in WS and DS, using tasks which tap both fundamental and more complex numerical skills, such as arithmetic.

Part Two: Empirical Studies

7.7 Introduction

This part of the chapter encompasses an investigation of some aspects of number performance in older children and adults with WS and DS. Their performance on number tasks is of interest because there have been many reports from parents about the difficulties their children face when dealing with number in everyday situations. This work is among the first to explore experimentally number difficulties in people with WS. Previous work has involved very few participants and has focused on different aspects of number competence. There has been a little more research into DS number, but the experiments reported below have never previously been conducted with participants from either of these two clinical populations.

The number domain is a large one and encompasses many different abilities. For the purposes of this thesis, it was necessary to focus on particular aspects of number. Two were chosen: fundamental number understanding and basic number skills. The fundamental aspect of number understanding task was the ability to make magnitude comparisons. Several researchers suggest that this ability corresponds to an understanding of number meaning. In other words, if an individual can compare the size of two Arabic numerals, it is likely that they have a fundamental understanding of the numerosity that they represent (Dehaene et al., 1990). This task is of particular interest in this thesis because it allows for the investigation of abilities in adulthood, the likely precursors of which can be measured in infancy. In Chapter 6, Experiment 1, the numerosity discrimination task taps number-relevant competencies in infants but in their very early stages of development. In the adult task, the semantics of number are tapped whereas in the infant task the infant must only discriminate a novel numerosity from a familiar one. However, both tasks involve processing two numerosities.

In Part One of this chapter, the mechanisms underlying number comparison were discussed and it was noted that children as young as 6 years exhibit the symbolic

distance effect. The first three experiments will examine the performance of adults and older children with WS and DS on tasks which normally elicit this effect, to ascertain whether it is present in these groups. If it is not, it is likely that number impairments present in more complex tasks could have their roots in the fundamental understanding of numerosity. If, however, normal effects are produced, then it is probable that a higher-level aspect of number or related cognitive processes may be impaired.

The fourth experiment is an investigation of the second aspect of number: basic number skills. These skills were assessed using a battery of number tasks, which cover several aspects of number from rote counting to basic operations. It is of interest to investigate whether performance on these measures reflects that seen in the fundamental number task, or whether these skills are separate. It is likely that many of them will call upon fundamental number understanding. The battery will allow a comparison to be made between the number skills of individuals with WS and DS for the first time. It is expected that the performance of both groups will be impaired. If mental age is a good indicator of number performance in these groups, it is likely that the performance of the atypical groups will be similar to that seen in the MA-matched group.

7.8 Experiments One and Two: distance effect and accuracy in dot comparison

7.8.1 Method

Participants

8 individuals with WS took part in these studies along with 9 individuals with DS, who were CA and MA- matched to the WS group, 8 typically developing children with CAs which match the mental age of the WS group on the BAS, and 8 individuals who were CA-matched to the WS group. However, only 7 people with DS completed the task, one pressed the same button continuously and would not attend to the screen another refused to do the task. The mean chronological age for each group, including

only those participants who completed the task, is reported in Table 7.1 below. In addition, the mean mental age from the BAS is reported.

Table 7.1 Mean chronological and mental ages for each group

	Mean CA (years)	range	Mean MA	range
WS	20;9	10;11-32;9	6;9	5;1-9;4
DS	24;3	11;4-35;3	5;9	5;1-6;4
MA	6;11	5;2-8;11	-	-
CA	21;1	9;10-29;8	-	-

Stimuli

Participants were presented with two arrays of dots simultaneously on a computer screen. Before the arrays appeared participants were alerted by a cross which appeared in the middle of the screen. The inter-stimulus interval was 995 ms and stimuli remained on the screen until the participant made her response. In experiment 1, the dots were presented in a random configuration, whereas in experiment 2 a canonical configuration was used. The canonical patterns were like those seen on dice or dominoes.

Design

Three aspects of the stimuli were varied in each experiment: (1) Participants were presented with a pair of arrays, which had numerosities that were either close together (a difference of 1, 2 or 3) or far apart (a difference of 5, 6 or 7). This was called the “split”; (2) Distance was also varied. This meant that the arrays differed in numerosity by 1, 2, 3, 5, 6 or 7; (3) Stimuli were presented with the larger numerosity on the right or with the larger numerosity on the left. Each pair of arrays was presented 4 times, and the order of presentation was varied for each participant. The distance was not used in the final analysis because there were different numbers of stimuli at each distance.

Procedure

Each participant was presented with a practice block before testing proper began. MA-matched controls and participants with DS and WS did a full practice block of 16 trials. CA controls were given three practice trials, in order to shorten the test session and because these participants could perform the task immediately. The test session consisted of 72 trials. Participants were told to respond to the stimuli with one of two keys which were highlighted with bright stickers on a computer keyboard (Z and M). They were told to depress the key on the same side as the array with the larger number of dots. If necessary, this was demonstrated during the practice block. These instructions were repeated when necessary. This occurred during most trials for the WS and DS groups. Encouragement was given throughout to keep participants on task, given the long duration of the session.

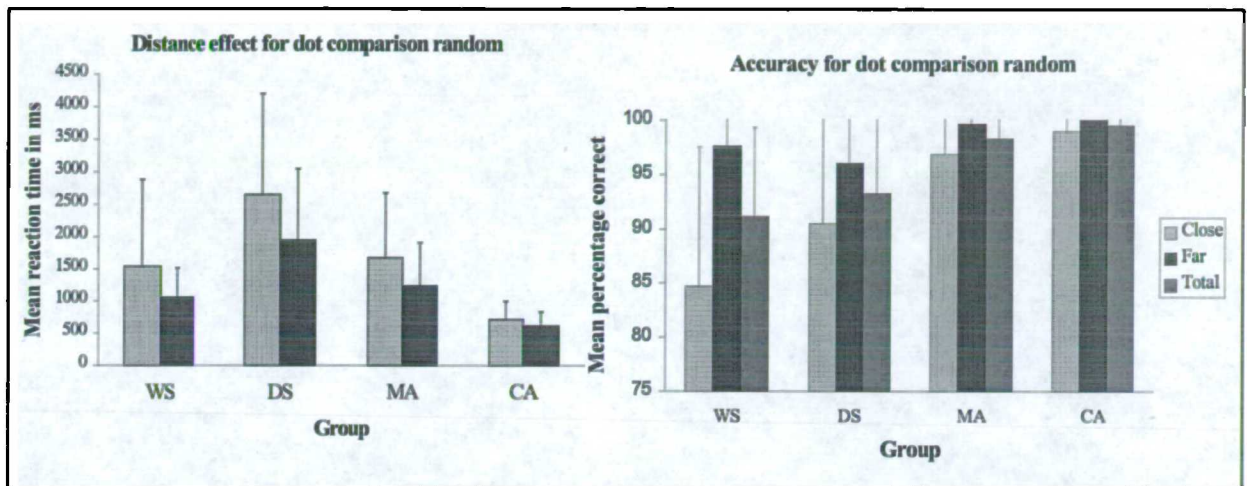
7.8.2 Results for Experiment One: dot comparison random

Each participant's results were sorted according to split, large or small, and then into blocks of four trials which were identical. Incorrect responses were removed and the median of each four trial block was calculated. Outliers for each participant were found using a box and whisker plot. The main body of the plot, represented values between the 25th and 75th percentile. Any reaction times which were more than 1.5 box lengths above or below these values were excluded from the analysis, according to the instructions provided in the SPSS statistical package (Kinnear & Gray, 1999). Outliers were removed in order to prevent the means from being skewed. Any extreme values are likely to be due not to numerosity comparison ability but to a lapse in attention. The percentage of trials omitted as outliers for each group is shown in Table 7.2. The mean reaction times and standard deviations for each group are presented in Figure 7.2, along with the percentage of trials correct. Readers interested in individual data for both reaction times and accuracy can find these tabulated in Appendices G and H respectively.

Table 7.2 Percentage of trials omitted as outliers by group for dot comparison random

Group	% outliers
WS	4.8
DS	5.6
MA	4.2
CA	7.9

Figure 7.2 Distance effect and accuracy for dot comparison random



Williams Syndrome group and Down's Syndrome group.

As illustrated above, the WS group's responses were faster than the DS group overall. T tests were carried out to investigate the difference in mean reaction times for trials with a large split or a small split. There was no significant difference in reaction times for the WS group, $t(df\ 7) = 1.42$, n.s. This means that the WS group did not exhibit a robust symbolic distance effect., although the results were in the correct direction. The DS group's responses did display the distance effect, $t(df\ 6) = 4.17$, $p < .01$.

Control groups.

A significant difference was found between the mean reaction time for stimuli with a large split and stimuli with a small split for the MA-matched children, $t(df\ 7) = 2.18$, $p < .05$. This was also true for the CA-matched group, $t(df\ 7) = 3.83$, $p < .01$.

Accuracy

Accuracy must also be considered for these tasks. The percentage of trials correct was compared across the four groups in two ways. First the percentage of total trials correct was analysed, then differences between close trials and far trials were examined. Heterogeneity of variance in these data meant that non-parametric statistics were used. A Kruskal-Wallis one way Anova revealed that there was a significant difference between groups in the total proportion of trials correct, chi-square = 7.99; df 3, $p < .05$. When the proportion of items correct was calculated separately for close pairs and far pairs, there was no effect of group for far pairs, chi-square = 2.06; df 3, n.s. For close pairs, there was an effect of group, chi-square = 8.36; df 3, $p < .05$. Post hoc tests did not reveal where this difference lay for close pairs or for the total percentage of items correct. However, the Kruskal-Wallis ranking indicated that the WS group performed worse than other groups for close pairs and total number of items correct. In addition to poor accuracy, the WS group does not exhibit a statistically significant distance effect, which indicates that these individuals may have a different basis for their discriminations.

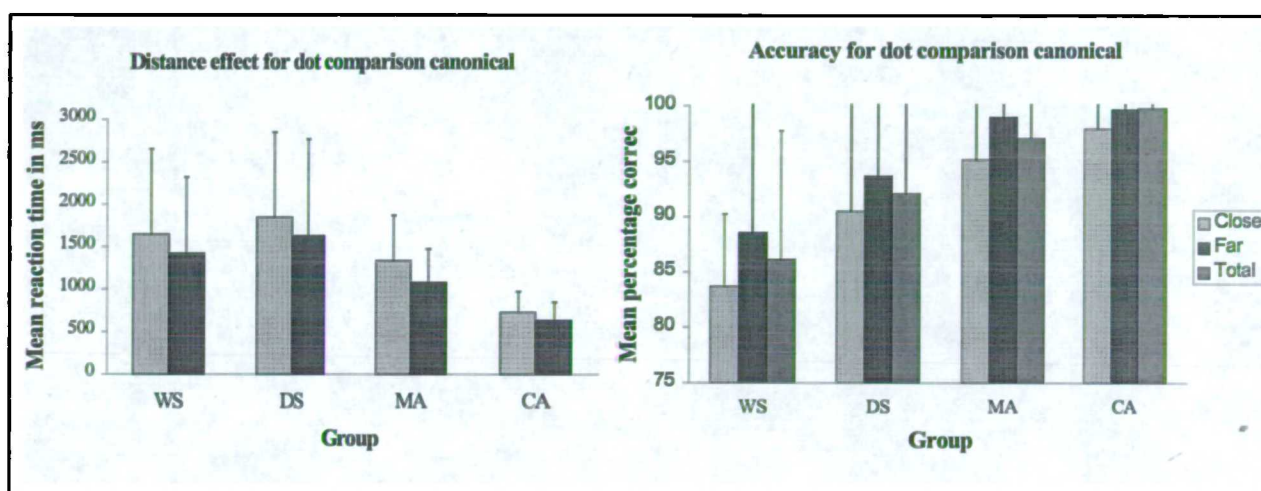
7.8.3 Results for Experiment Two: dot comparison canonical

Each participant's results were sorted according to split, large or small and then into blocks of four trials which were identical. Incorrect responses were removed and the median of each four trial block was calculated. Again, outliers were removed from each individual's data. The percentage of trials omitted as outliers for each group is shown in Table 7.3. The mean reaction times and standard deviations for each group are presented in Figure 7.3 along with the percentage of trials correct. Readers interested in individual data for both reaction times and accuracy can find these tabulated in Appendices G and H respectively.

Table 7.3 Percentage of trials omitted as outliers by group for dot comparison canonical

Group	% outliers
WS	8.0
DS	3.5
MA	6.3
CA	7.1

Figure 7.3 Distance effect and accuracy for dot comparison canonical



Williams Syndrome group and Down's Syndrome group

As for the experiment in which random dot arrays were used, although the responses of WS group were in the correct direction, they did not display a robust distance effect, whereas those of the DS group did, $t(df\ 7) = 1.44$, n.s. and $t(df\ 6) = 4.05$, $p < .05$.

Control groups

Again, both the MA and CA- control groups exhibited a robust distance effect. Their were significant differences between response times to close and far stimuli, $t(df\ 7) = 2.87$, $p < .01$ and $t(df\ 7) = 3.74$, $p < .01$ respectively. Response times to stimuli that were far apart were faster than those to stimuli that were close together, as would be expected. If numbers are close together there is more interference in the discrimination process, so participants are slowed down.

Accuracy

Accuracy was analysed for all four groups using the same procedures as for dot comparison canonical. A Kruskal-Wallis one way Anova revealed that there was an effect of group on accuracy for the total number of items, chi-square = 11.85; df 3, $p < .01$. However, post hoc tests revealed no significant difference between any pair of groups. This might be due to the presence of an overall difference, but no significant differences between single pairs of groups, e.g., WS versus DS. When the percentage of trials correct for close pairs and far pairs was compared for each group, a significant effect of group was found for close pairs, but not far pairs, chi-square = 12.53; df 3, $p < .01$ and chi-square = 2.06; df 3, n.s. This indicates that the close trials were harder than the far trials as would be expected. A Tamhane post hoc test revealed that the WS group made fewer correct responses than the MA- or CA-matched groups, $p < .05$. This suggests the WS group had difficulty with this task and that perhaps their numerosity representations were not as clear as those of the control groups. However, the fact that the individuals with WS performed particularly poorly on close trials suggests that they have some underlying semantic representation. This is because a semantic representation is likely to be at the root of the distance effect (Dehaene et al., 1990), with a small difference in the two numerosities causing interference in the discrimination task. It could be that these numerosity representations are not as well-defined in WS as in other groups.

7.9 Experiment Three: Arabic numeral comparison

7.9.1 Method

Participants

All the participants from experiments 1 and 2 took part in experiment 3, with one additional individual with DS, who refused experiments 1 and 2. The mean CA for the group of 8 individuals with DS was 22;5 years (range 9;11- 35;3) and the mean MA was 5;9 (range 5;1-6;4).

Stimuli and procedure

This procedure is the most commonly used when testing numerosity comparisons with adults. The stimuli for Experiment 3 were the Arabic numeral equivalents of those used in experiments 1 and 2. The numerals were presented in 48 point bold Arial font. All participants were able to read the numerals 1-9. For this experiment, the inter-stimulus interval was 994 ms. The procedure was the same as that used in experiments 1 and 2. The participant was asked to respond to the larger of the two numbers.

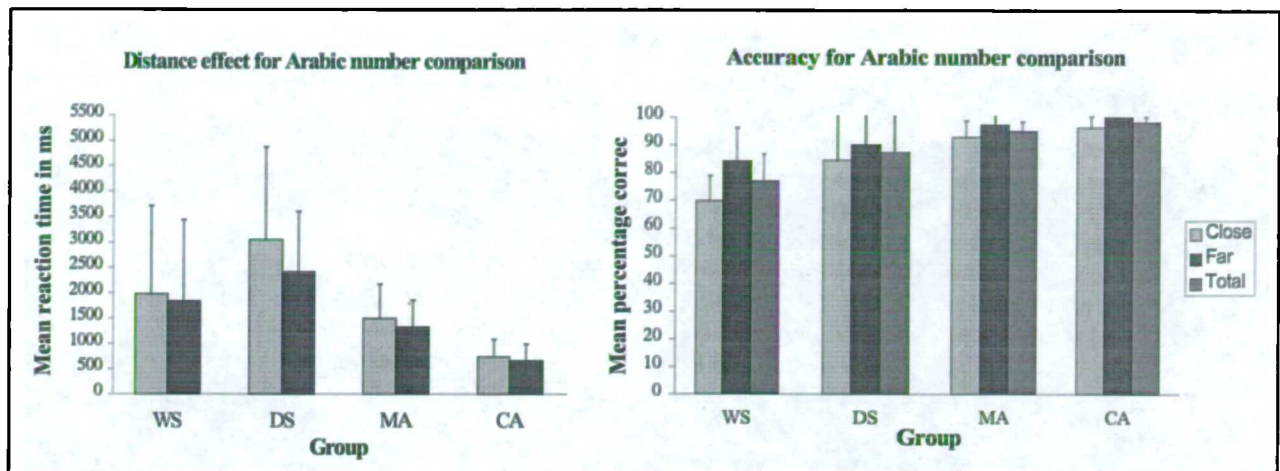
7.9.2 Results for Experiment Three: Arabic numeral comparison

Each participant's results were sorted according to split, large or small, and then into blocks of four trials which were identical. Incorrect responses were removed and the median of each four trial block was calculated. Outliers were also removed, as in Experiments 1 and 2. The percentage of trials omitted as outliers for each group is shown in Table 7.4. It was predicted that if number meaning was comprehended, a distance effect would be present in the data, with reaction times to the large split being faster than those for the small split. The mean reaction times and standard deviations for each group are presented in Figure 7.4 along with the percentage of trials correct. Readers interested in individual data for both reaction times and accuracy can find these tabulated in Appendices G and H respectively.

Table 7.4 Percentage of trials omitted as outliers by group for Arabic numeral comparison

Group	% outliers
WS	5.6
DS	6.3
MA	3.5
CA	4.8

Figure 7.4 Distance effect and accuracy for Arabic numeral comparison



Williams Syndrome group and Down's Syndrome group.

Again, no distance effect was seen in the reaction times from the WS group, $t(df\ 7) = 1.44$, n.s. In contrast to data from the dot experiments, the data from the DS group for Arabic numeral comparison, while exhibiting a trend towards the distance effect, was not significant, $t(df\ 7) = 1.63$, $p < .07$. This may be due to the higher standard deviation in the data for this task (869 versus 392 for dot comparison canonical) or a characteristic of the task, as discussed below. (A power calculation revealed that in order to achieve a significance level of 95% with a power of 80%, 30 participants with DS would need to be tested. However, this is heavily influenced by the large variance in the data caused by extremely high reaction times from one participant. If this participant is removed from the power calculation, then only 21 participants are required by the analysis.)

Control groups

As expected, both the CA and MA- matched groups displayed the distance effect. For the MA- matched group the mean reaction times for far pairs and close pairs were 668 ms and 540 ms respectively. This difference was significant, $t(df\ 7) = 2.62$, $p < .01$. The difference for the CA- matched group was between a mean reaction time of 725 ms for close pairs and 650 ms for far pairs, $t(df\ 7) = 3.71$, $p < .01$.

Accuracy

Again, accuracy was measured using the percentage of items correct and the data were analysed as in Experiments 1 and 2. A Kruskal-Wallis one way Anova indicated that there was a significant effect of group on the percentage of the total items correct, $\chi^2 = 15.36$; $df\ 3$, $p < .01$. A post hoc test revealed that accuracy for Arabic numeral discrimination in the WS group differed significantly from the MA- and CA-matched groups, Tamhane, $p < .05$. When the percentage of items correct was considered separately for close trials and far trials, there was an effect of group for both close trials ($\chi^2 = 14.04$; $df\ 3$, $p < .01$) and far trials ($\chi^2 = 16.39$; $df\ 3$, $p < .01$). Post hoc tests indicated that the differences lay between the WS and MA-matched group and the WS and CA-matched group for close pairs, Tamhane, $p < .05$. For far pairs, post hoc tests indicated the difference was between the WS and CA groups, Tamhane, $p < .05$.

In sum, then, for Arabic numeral comparison the WS group continued to show no robust distance effect. However this was also true of the DS group but this could be due to the variance in the data or perhaps to the greater demands of this task, as explained in the discussion. The WS group made significantly more errors in total than either of the control groups, suggesting that there is likely to be some abnormality in number processing in this group.

7.10 Discussion for number comparison studies: Experiments One, Two and Three

The participants in the WS group behaved differently from the other groups. They did not exhibit a robust distance effect. In other words, they did not take significantly longer to discriminate between arrays which have similar numerosities, e.g., 2 and 3 and those which are very different, e.g., 2 and 6. This was true for random and canonical dot comparison as well as for displays with Arabic numerals. All other groups displayed the distance effect, which is a very robust phenomenon, for dot comparison. However, for Arabic numeral comparison the difference between reaction times for close and far stimuli was not significant for the DS group. This

finding is reflected in the accuracy of the DS group for Arabic numeral comparison. Participants with DS made more errors in the Arabic numeral experiment than in the other two. It is likely that this task was more demanding than the dot comparison because the task involved two steps. In order to compare the numerals, participants first had to transform them into numerosities, whereas for the dot comparison the displays were already in magnitude form.

With respect to accuracy, the WS group was the least accurate of all groups for both the Arabic numeral comparison and dots canonical experiments. However, it should be said, all groups perform well above chance. No clear post hoc differences between groups were found for accuracy with the random dots, although individuals with WS were the worst performers. However, for all the experiments, the performance of the WS group was above chance level, so participants with WS could do the task. The accuracy of both the MA-matched and CA-matched groups was high. Accuracy was generally lower for close pairs than far pairs. This would be expected because as discrimination becomes more difficult, more errors are made, in addition to an increase in reaction time.

Analysis revealed that the WS group performed worse than the control groups in all three tasks. They make more errors when the arrays are close in number, so the distance effect is to some extent reflected in their errors. This suggests that although members of WS group do not exhibit a difference in reaction time, they have some understanding of numerosity. However, their performance is still anomalous. Indeed, some of the subjects even show a trend towards an inverse distance effect, with close trials exhibiting faster reaction times than far trials.

In my view, the results from the WS group suggest that there are anomalies in their basic numerical processing. The distance effect is a very robust indicator of the presence of a clear mental representation of numerosity, and this may be different or weaker in individuals with WS. The distance effect is apparent with the dot stimuli in the DS group, who are matched on mental age with participants with WS, but behave

like their typically developing counterparts. This result suggests that the impaired performance of the WS group is not due to general cognitive impairment.

It may be that a fundamental difference in the representation of quantity in the WS group is at the root of later, higher-level number difficulties. The results from the experiments described above are particularly striking, given the results from the WS group in infancy. Recall that in infancy the WS dishabituated to a novel numerosity (3) after familiarisation with arrays of 2 objects. It is possible that the individuals with WS have an intact representation of small numerosities, and may well be able to discriminate a change in numerosity, but perhaps cannot represent the precise nature of the change. By contrast, the DS group was unable to detect the change in infancy but perform better on the adult task. This may constitute a delay in the maturity of numerosity discrimination in DS infants, leading to delayed numerosity understanding in the DS group. However, it is likely that individuals with DS use the same processes for number understanding as normals, whereas the WS group may be relying on different processes.

If the magnitude representation, or mental number line, were normal in individuals with WS, then one would expect a typical symbolic distance effect. Its absence or weakness suggests that number-related representations made by these individuals are different. For example, it is possible that the WS group represents number such that magnitudes do not have regular distances between them, so that the interference effects found in other groups are different in the WS group. Of course the type of representations used by the WS group are, as yet, unknown. Data from the number battery, which is presented below, may shed further light on the properties of numerical representations in WS and DS.

7.11 Future directions for numerosity discrimination

The numerosity discrimination tasks yielded some very interesting results. However, in future research, the number of trials in these experiments could be reduced. It was extremely difficult for individuals with WS and DS to remain on task for all 72 trials.

Since it is possible to demonstrate the distance effect with fewer trials (Sekuler & Mierkiewicz, 1977), this should be done.

As is often the case with research into atypical development, there were problems with heterogeneity of variance in the data. This meant that non-parametric statistics had to be used. These statistics can be less rigorous than parametric equivalents and may in some cases lack the power to reveal differences. There were two occasions where despite revealing a difference between all the groups, statistics were unable to pinpoint where the difference resided. A larger number of subjects might ameliorate some of these difficulties by providing statistical power.

7.12 Experiment Four: Number Battery

In addition to the computer-based number tasks, the participants completed a number battery. This was designed for use with neuropsychological patients (Girelli, 1999). It comprises: number seriation and dot seriation, rote counting, Arabic numeral reading, matching dots to Arabic numerals, “what comes next/before?” and basic operations with single digits (addition, subtraction and multiplication). The multiplication task involved picking the correct response from 3 possible responses: one correct, one wrong and not in the table (e.g., $2 \times 4 = 9$) and one which appears in the table of one of the numbers (e.g., $2 \times 4 = 6$).

7.12.1 Methods

Participants

The participants in the WS, CA-matched and MA-matched groups were the same as those in Experiments 1 and 2. In the DS group, 2 individuals could not complete the battery, one could not count to ten, and the other refused to take part. The mean chronological age of the 6 participants from the DS group was 26;4 years, range 17;11-35;3. The mean MA on the BAS II was 5;10 years, range 5;7-6;4.

Procedure

The battery was administered to each participant individually. A description of the tasks is provided in Table 7.5. Responses were given either orally, by pointing to the correct response (matching and multiplication) or by putting cards into the correct order (seriation). Some individuals were given paper in order to complete the arithmetic tasks. Participants completed the items on their own, but were given encouragement or hints if they did not know what to do. On several occasions an example was given, in order to clarify the task. Where necessary, for motivational reasons or time constraints, a shorter version of the Arabic seriation task was used. For CA- and MA-matched groups, if the participant could complete the last three items, then earlier items were not tested and their performance was deemed 100% correct. The last item of dot seriation was used in the same way. If performance was not 100% correct, all items were administered. Any other deviations from the battery are noted in the tables 7.6 or 7.7.

7.12.2 Results and discussion

The group data are for all those individuals who attempted the complete task. Data are also presented for each individual. This is done because some individuals did not complete all the items for a particular task. They may have only attempted a limited set of items, so their performance could not be compared with that of other participants who attempted every item.

Each participant's performance is reported as the percentage of items correct for each task in Tables 7.6 and 7.7. These data highlight several interesting findings. First, the majority of the MA and CA-controls performed at or near ceiling. The rote counting performance of the MA group was less good above the number 20, but it must be noted that those children who had problems were younger than 6 years old. This was also reflected in the seriation task, where the youngest MA-matched children only managed to put single digits into the correct order. The performance of MA-matched controls on the arithmetic tasks was mixed. Several individuals did not attempt the multiplication tasks. It should be noted that according to the UK National Curriculum,

these children would not be expected to complete all the multiplication tasks. In general, the MA group could do the addition task, at least to their ability level, and 5 were successful with subtraction. The CA-matched group reached 100% performance on all but two tasks: matching and multiplication. It would be expected that matching would not be perfect, unless participants were counting the dots instead of using a subitizing strategy. It is easy to make errors when judging numerosity of arrays of 9 or 10 dots randomly arranged.

Table 7.5 Tasks in the number battery

Task	Example
Arabic numeral seriation (15 items) Put numbers in serial order in order.	6, 3, 1, 8 20, 6, 9, 38 9, 25, 17, 112
Dot seriation (6 items)	Dots vary in size. An example sequence: 5,2,3,1
What comes next/before? (14 items each) Experimenter says x, participant says what comes next/before.	E.g. 1 - 11- 65- 17- 10-
Matching (20 items - 10 random/10 canonical) Point to the Arabic numeral which matches the dot array. One option is correct, one is incorrect but close, the other incorrect but far.	Array of 5 random dots. Choice of : 2, 6, 5 Array of 3 dots in a canonical pattern Choice of : 6, 4, 3
Reading Arabic numerals (22 items) 9 single digit items 11 double digit items 3 triple digit items	4 3 11 80 100 250
Rote counting	forward 1-20 forward 25-35 backwards 20-1
Addition (25 items possible) Presented on cards (read out if necessary) verbal response	2+3 6+1 8+8 (maximum sum 16)
Subtraction (as above- 25 items)	2-1 7-0 9-4
Multiplication Alternative answers provided: One correct , one incorrect in the table, one incorrect not in the table.	2x4 = 8 (correct) 6 (in either 2 or 4 times table) 9 (not in table)

Second, as would be expected, the performance of the WS and DS groups was poorer than the performance of typically developing counterparts, but more interestingly there were marked differences between the clinical groups. The WS group had more difficulty with the tasks than the DS group. In order to compare the performance of the WS and DS groups statistically, a t-test was carried out with the average performance on the following tasks as the dependent variable: counting 1-20, What comes next? Seriation (dots and Arabic), reading (ones and others) and matching. Other tasks were excluded from the analysis because percentages could not be calculated from the data collected. The performance of the DS group was better overall than the WS group's, $t(df\ 6) = 2.34, p < .05$.

The greatest differences in performance between the WS and DS groups were for the "What comes next/before?" tasks and Arabic and dot seriation. Many of the WS group had difficulty with the "What comes before?" task for two possible reasons: (1) they had difficulty understanding the instructions and often offered the next number in the sequence, even when reminded not to, and (2) because they could not resort to their rote count so readily for this task. They were unable to merely count on.

There were several types of errors from participants with WS. Some involved saying what came before in a "next" task, and others were similar to those seen in patients (Dehaene et al., 1991). The majority was with double-digit numbers, e.g. skipping from 40 to 50 or from 11 to 20. This suggests that the number line in these individuals with WS may not be well specified. Some of the errors made by participants with WS involved changing the class of a number. Numbers are divided up into classes with ones, teens and then multipliers of tens, hundreds and thousands. Individuals with WS swapped between classes when providing the next number in a sequence. Instead of saying 14 comes before 15 the response would preserve some of the lexical nature of the correct number, so 4 would be part of the response but instead of being part of the teens (14) it would be changed to part of the tens (48) and an extra lexical entry would be added. In another response, 2 would be moved from the teens (12) to the tens (20). These responses and others like them again suggest that numerosity is not clearly specified in WS. It is important to note that these kinds of errors were not present in

the responses from the DS group. The class or size of the numbers was preserved in their responses, e.g. 30-40 or 40-50 but the lexical elements were changed (e.g., 31-33). This type of error was also present in WS responses, but was in addition to the out of class errors mentioned above.

For the seriation task, problems often occurred when participants misread Arabic numerals, so mistook 17 for 71, for example. This is known as an inversion error which is often made by young children and is a sign of immature numerical representation. Further examples of this were found in the data from the reading task. In the reading task, several of the individuals with WS were successful with single digits but their performance deteriorated with double digits, mirroring the findings from the “What comes next/before” task. This pattern of responses might reflect a problem with the syntax of number. Syntactic errors are reflected in the structure of numbers. “30” can be thought of as 3 tens, or 12 as the 2nd of the teens. Errors made by individuals with WS changed the syntax of numbers either by inversion from 14-41 or by incorporating other syntactic structures, e.g., 250- 2000. These errors suggest the individuals with WS have trouble switching from one numerical code to another. In this case, the problem is switching from written Arabic codes to spoken numbers. This conversion between the Arabic code and the phonological speech code is likely to involve two steps. The first is a conversion from Arabic code to numerosity and the second from the numerosity to phonological output. Further research is necessary to pinpoint where precisely the impairments in WS are centred. One or both of these pathways could be entailed. Alternatively, people with WS may bypass semantics or the numerosity code altogether. Interestingly, weak semantics characterises their language and reading too (Laing et al., submitted). For now, it is sufficient to note that there are problems in the transcoding process and that these differ from those in individuals with DS.

Other errors are of less interest, but will be mentioned briefly. Where errors occurred in the matching task, these were to a sensible alternative on all but one occasion. If the number of dots was 5, given a choice of 2, 6, or 5, the participant would choose 6. The WS group was a little better at this task than the DS group. This was due to the

participants with WS resorting to an exact counting strategy more frequently than the other groups, whose members more often gave immediate responses, probably based on a form of subitizing.

The data from the fundamental number tasks turn out to be very interesting. Unfortunately, it is difficult to make a fair assessment of the addition, subtraction and multiplication data. Many of the participants with WS and DS could not complete a full set of each type of sum. Certainly, there was more awareness of addition and subtraction processes than multiplication, but the participants in the WS group had weaker knowledge of number bonds and resorted to fingers and dots on a page more frequently than the DS group. Indeed, 2 of the people with WS did not even seem aware of the underlying concepts of addition and subtraction. They had to be told by the experimenter how these operations worked. Their attempts were not scored, because they did not constitute true addition or subtraction. Instead, they constituted drawing sets of circles and counting them, for example. This may be due in part to the differences in educational opportunities available for each group. These differences are an important factor when considering achievement in formal arithmetic (Nye et al., 1995).

The lack of an equal number of trials for all the groups is an unavoidable shortcoming of this part of the study. However, it was often very difficult to motivate participants to attempt the arithmetic, especially at the end of a long test session. These were also the items that all participants found most challenging and people with learning difficulties often have avoidance strategies for tasks they know will be difficult.

Table 7.6 Percentage of items correct on the number battery by individual: WS and DS

Number battery WS										DS									
	JN	TS	LS	AD	CC	GM	RD	NM	Ave	SD	EB	CM	AB	DT	SH	JP	Ave	SD	
Counting 1-20	85	100	100	100	100	100	100	100	98.13	5.30	100	100	100	100	100	100	100	0	
Counting 25-35	*	100	upto28	100	upto34	100	100	prob			100	100	100	100	100	100	100	0	
Backwards 20-1	21.05	100	*	*	*	*	50	prob			100	100	50	*	100	100	90		
Next	50	71	64	100	86	100	64	92.86	78.48	18.80	100	100	92.86	100	100	85.71	94.43	5.98	
Before	*	100	64	92.8	0.00	92.86	50	79	68.38	34.23	100	100	*	*	100	100			
Seriation dots	83	80	50	80	40	60	67	83	67.88	16.52	100	100	100	100	100	100	100	0	
Seriation Arabic	80	100	33	100	47	100	67	100	78.38	26.85	100	100	100	100	100	100	100	0	
Reading Arabic	89	100	100	100	100	100	100	100	98.63	3.89	100	100	100	100	100	100	100	0	
1s																			
Others	36	100	85	100	85	100	92	100	87.25	21.74	100	100	100	100	100	100	100	0	
Matching	100	80	70	100	85	95	75	95	87.5	11.65	90	90	80	65	80	90	82.5	9.87	
Add	*	6/11	dot counting	80	72	88	36	84			84	80	*	4/15	9/10	10/10			
Sub	*	8/9	60	84	56	9/10	24	32			76	92	*	3/8	7/13	7/13			
Mult	*	2/7	*	6/15	*	13/16	6/13	*			64	*	*	*	*	*			

* Not done

Table 7.7 Percentage of items correct on the number battery by individual: MA- and CA-matched groups

Number battery		MA																CA							
		BB	HH	GB	CM	SW	JA	TI	JR	Ave	SD	VW	GS	RP	ZS	CE	CB	GP	RM	Ave	SD				
Counting 1-20	100	100	100	100	100	100	100	100	100	100	0	100	100	100	100	100	100	100	100	100	0				
Counting 25-35	0	0	100	100	100	100	100	100	100	75	43.30	100	100	100	100	100	100	100	100	100	100	0			
Backwards 20-150	100	100	100	100	100	100	100	100	100	93.75	16.54	100	100	100	100	100	100	100	100	100	100	0			
Next	79	100	93.33	100	100	100	100	100	100	96.54	6.98	100	100	100	100	100	100	100	100	100	100	0			
Before	28.6	100	100	100	92.86	100	100	100	100	90.18	23.39	100	100	100	100	100	100	100	100	100	100	0			
Seriation dots	100	100	100	100	100	100	100	100	100	100	0	100	100	100	100	100	100	100	100	100	100	0			
Seriation Arabic 20	20	100	100	100	100	100	100	100	100	80	34.64	100	100	100	100	100	100	100	100	100	100	0			
Reading Arabic 1s	100	100	100	100	100	100	100	100	100	100	0	100	100	100	100	100	100	100	100	100	100	0			
Others	92.3	100	100	100	100	100	100	100	100	99.04	2.55	100	100	100	100	100	100	100	100	100	100	0			
Matching	85	95	100	95	85	100	100	100	95	94.38	5.83	100	95	100	95	100	100	100	100	100	98.57	2.26			
Add	**	100	76	92	100	100	100	100	100			100	100	100	100	100	100	100	100	100					
Sub	**	8/12	100	**	96	100	100	80	100			100	100	100	100	100	100	100	100	100					
Mult	**	**	**	**	**	**	**	**	100			92	100	100	100	96	100	100	100	100					

** Not yet done in school

7.12.3 Future research

In future research, a different number battery might be more usefully employed. The length of some of the tasks in my study meant that it was difficult to keep participants engaged. The arithmetic tasks turned out to be too demanding to produce interesting results. A battery such as that developed by Baroody (1986), along with tasks based on items from the Teacher Questionnaire (Sloper et al., 1990), might have been much more enjoyable for the participants and have yielded data which were just as informative but more plentiful. It would be useful to present a scaled set of arithmetic tasks to the participants and to investigate whether they used different strategies according to task demands. Perhaps, as happens with younger typically developing children, the groups would be able to retrieve the answers to easy problems from memory but would resort to other strategies for harder problems. The more difficult problems might highlight the type of strategies used by each group. However, despite its shortcomings, the battery in the present experiment did allow for one of the first comprehensive assessments of the number abilities of individuals with WS and a comparison with a DS group. The results suggest that the WS group have subtle problems with number across several tasks which are more serious than those exhibited in the DS group.

Now that a general assessment of performance has been carried out, it will in future be possible to look more closely at particular aspects of number processing and manipulation. Unpacking the links between language ability and number would be particularly fruitful, given the interesting language profile in WS. It might also be useful to test them using tasks which would add to the body of data which are used to decide between different models of number processing. These would need to tap separately, as far as possible, the different number codes: Arabic, verbal-spoken and written and quantity (e.g., dots). The seeming lack of a clear numerosity representation in the WS group may shed light on how this representation is used in the normally functioning number system. The tasks which are most impaired in the WS group seem to be those that rely most heavily on a clear mental representation of

numerosity, i.e., on the semantics of number. Difficulties with the syntax of number are also prevalent.

General Discussion and Chapter Summary

The results of the computer-based numerosity comparison tasks in experiments 1, 2 and 3 highlight real abnormalities in the processing of number by individuals with WS. The WS group was the only one which did not exhibit a reliable symbolic distance effect. This was true for random and canonical dot arrays. Both the WS and DS groups had difficulty with the Arabic numeral task, but the distance effect was approaching significance in the DS group. These results suggest that while the other groups have a normal understanding of numerosity, this is not true for the individuals with WS. The data from the number battery paint a similar picture of impairment in WS. Although individuals with WS appeared to be able to count by rote, at least to 20, problems emerged as soon as they had to manipulate numbers and not merely reproduce a verbal string. This was clear even when individuals were asked to put numbers in the correct order. Individuals with WS seemed to have difficulty with the syntax of number, and thus became confused when switching from 10 to 11 or 20 to 21, for example. These problems with ordering number suggest that representations of magnitude may not be well linked to their respective symbols.

It is nonetheless to be noted that although they had difficulty with tasks in the number battery, individuals with WS can abide by counting principles. However, it is noteworthy that the tasks in which they are successful usually require small numerosities. It may therefore be possible to rely on another system to implement the counting principles correctly. However, with larger numbers which appear in the number battery, a clear understanding of the link between numerosity and symbol is necessary (Dehaene et al., 1990) and it seems absent in the WS group.

The unusual data from the WS group highlight another important aspect of the study of atypical development. A fundamental difference in processing such as that seen in

WS number comparison may have varying effects across other aspects of the domain. So, for small numbers, the effect of the impairment can be by-passed in every day functioning, but for larger numbers and more complicated tasks, this may not be possible. For more complex tasks, the deficit exhibited in the number comparison task might be present, but in a different guise. So for example, the problem with linking number meaning or numerosity to symbols might underlie the difficulties that the WS group had with the reading or seriation tasks.

In sum, then, the impairments in number in adults and older children with WS are different from those seen in the DS group. It is likely that the performance of individuals with DS is immature, as evidenced by their slow reaction times, but follows the typical pathway. In contrast, individuals with WS appear to deviate from normal behaviour right from the very core abilities of number. This basic difference leads to a cascade of further difficulties.

Both numerical and language abilities in Williams Syndrome and Down's Syndrome have now been considered. In the final chapter, the principal findings of this thesis will be summarised and their importance for both theoretical considerations and future research will be discussed.

8

Summary & Conclusions

My thesis aimed to test whether the neuropsychological model of adult brain injury could be applied to neurodevelopmental disorders. To this end, the studies presented in this thesis were designed to investigate the infant phenotype of Williams Syndrome and Down's Syndrome on tasks tapping aspects of language and number ability, and to compare this performance with that present in the mature phenotype.

It is crucial to examine cognitive ability in infancy, to investigate whether differences in performance across syndromes occur as a product of different developmental processes, affecting learning and the processing of inputs from the environment, or whether the differences in neurodevelopmental disorders are already present in infancy. In order to begin to apply a developmental approach, two aspects of cognitive development in WS and DS were examined both in infancy and in the phenotypic outcome, although of course in long-term postdoctoral research it will be essential to chart the full developmental trajectory between infancy and adulthood. The domains of number and language were chosen because language abilities differ substantially in the two syndromes in the mature phenotype, but number abilities are reported to be seriously impaired in both clinical groups. This difference in outcomes, both within and across syndromes, allows a number of important theoretical issues to be addressed.

Williams Syndrome and Down's Syndrome were chosen for study because they exhibit distinctive cognitive profiles in later life, and allow one to investigate whether the within- and between-syndrome differences are present in the infant starting state.

The results from 13 studies provide strong evidence that the assumptions of the adult neuropsychological model are indeed inappropriate for understanding developmental disorders. Over both the language and number domains, and even in general cognitive skills, the pattern of abilities present in infancy cannot simply be inferred from those seen in the steady state of middle childhood and beyond.

The following sections provide a summary of the results from these studies.

8.1 General cognitive ability in Williams Syndrome and Down's Syndrome

8.1.1 The infant cognitive profile

In order to examine the change in the cognitive profiles of the WS and DS groups over development, participants were first examined using standardised ability tests. Infants were tested with the Bayley Scales of Infant Development II (Bayley, 1993). In order to see whether the pattern of language and non-language abilities reported in adults with WS and DS was already present in infancy, the infants' performance on language and non-language items was compared. This was done by calculating the percentage correct for language and cognitive items for each infant. The division between cognitive and language items was taken from the facets described in the manual. Overall, the infants with WS performed better than the infants with DS. In addition, the specific patterns of results across the groups differed. In contrast to the adult data, infants with WS showed no significant difference between language and cognitive scores, whereas there was a significant difference for the DS group, with the latter being at a particular disadvantage on language items. Data from the younger, MA-matched controls shows a similar pattern. This suggests that infants with DS are exhibiting delayed development, similar to that seen in normal infants at an equivalent developmental level. By contrast, those with WS appear to display a deviant developmental pattern of results on the Bayley compared to the pattern in typical development.

8.1.2 The mature cognitive profile

The mature cognitive profile in DS and WS was also assessed to examine whether the pattern present in the infant data was reflected in adult performance. Two groups of older children and adults with DS and WS were assessed using the British Ability Scales. This was done to examine whether my group of participants conformed to the Williams Syndrome Cognitive Profile, as set out by Mervis et al. (1999) and, more importantly, to examine whether the profile found in the mature phenotype was the same as that found in infancy. The individuals in my sample did not conform to Mervis et al.'s criteria for the WSCP. It appears that a "classic" profile of WS, at least as operationalised by Mervis and her colleagues, is not seen in all individuals with this condition. That said, if results are analysed in a less stringent fashion, investigating the degree of fit instead of absolute conformity, then individuals with WS can be placed along a continuum, from a very strong fit to the classic profile to a rather less well-defined WS cognitive profile. It is clear that the WSCP is distinctive. None of the DS group conformed to the Mervis criteria and their degree of fit was also very low. Thus, the WS profile is not just a product of general retardation but is likely to be syndrome specific.

The results also suggested that the profiles of infants with WS and DS differ to a lesser degree than those exhibited in middle childhood and adulthood. Therefore, the different developmental trajectories followed by these syndromes must be responsible for the cognitive profiles seen in the steady state. This highlights the importance of characterising the infant state. If the infant cognitive profile were simply assumed from that present in adulthood, without empirical verification, then the subtle changes that occur over development would be missed.

8.2 Specific abilities in Williams Syndrome and Down's Syndrome: Language in the infant and mature phenotype

Previous research has shown that, in general, language is a relative strength in WS but a relative weakness in DS (Bellugi et al., 1994; Mervis et al., 1999). In particular vocabulary is relatively spared in individuals with WS when compared with their DS

counterparts. The studies in my thesis were conducted to investigate whether the pattern of vocabulary abilities in adults with WS and DS reflected that displayed in infancy, or whether differences in ability in the mature phenotype are the product of different developmental trajectories in each group.

8.2.1 Language in the infant phenotype

Vocabulary development in infants was assessed using an experimental method, preferential looking, and a parental report questionnaire, the MacArthur Communicative Development Inventory - CDI (Fenson et al., 1993). It was predicted that if infant performance could be inferred from the mature cognitive profile, then the infants with WS would perform better on vocabulary measures and more like CA controls than the DS group, who should perform like MA-controls. The data show that all groups could do the task. However, the relative pattern of looking time to matching and non-matching stimuli differed across the groups. The pattern for the WS and DS resembled that for the MA-matched group, suggesting that both groups were delayed in infancy, even though adults with WS have significantly better vocabularies than those with DS.

Data from the second study, using the CDI, revealed a similar pattern across the WS and DS groups. Parents were asked to complete the infant version of the questionnaire because the infants in my study had a mean mental age of approximately 15 months. These infants were above the chronological age on which this measure was normed and therefore they could not be given a standard score. Instead, the percentage of items marked as comprehended and the percentage of items produced were calculated. The difference between the comprehension and production level of each group was assessed. The data indicated that although all groups showed some advantage for comprehension over production, as would be expected in typical development, this difference was due to the particularly low level of production found in the DS group.

The higher level of production present in the WS group may be a first sign of the verbal advantage that they exhibit in later life. This small difference in the beginnings

of language acquisition between WS and DS could lead to marked differences later in development for a number of reasons. It is plausible that the fact that infants with WS produce more language leads to more language input from caregivers, because they see that the child is sensitive to their earlier input. This increased input could contribute in turn to further language development. For example, it is known that lexical development is important for the development of syntax (e.g., Bates, Bretherton & Snyder, 1988), so a larger lexicon in infants with WS could provide a better basis for later syntactic development. An alternative reason for the verbal advantage in WS is that the infants with WS may have a particular propensity to attend to and respond to language input, which might become more marked with development. For example, the way in which the WS brain develops may favour the processing of language stimuli over other input because its particular architecture is especially suited to, say, sequential stimuli. This bias may become more marked over time and so lead to greater differences between WS and DS as a function of development. Both increased input and a particular processing bias could also work in concert, leading with time to the WS advantage.

8.2.2 Language in the mature phenotype

In contrast to the infant data, the data from language studies conducted with older children and adults show that performance is better in WS than in DS. Receptive vocabulary performance was compared across the WS and DS groups, using the British Picture Vocabulary Scales (BPVS). The groups were matched according to CA and overall cognitive ability. Despite this, individuals with DS achieved lower age-equivalent scores than their WS counterparts. Older participants were also assessed on two aspects of syntactic comprehension: sensitivity to word order and to subcategorisation violations. The results indicated that for subcategorisation and word order, the WS group was more sensitive to violations than the DS group, producing more corrected sentences and acceptable responses to ungrammatical sentences. However, the WS group performed worse than CA controls. Overall, the results suggest that both groups are impaired in syntactic understanding, but that the impairment is greater in DS.

8.2.3 Summary

To summarise, despite similar delays in receptive vocabulary in infancy, individuals with WS and DS exhibit different language profiles in adulthood. While syntax remains a problem for both groups, the receptive vocabulary performance of individuals with WS is markedly better than that for DS. If one were to guess at the infant profile simply from the adult data, a very different and misleading picture would have emerged.

8.3 Specific abilities in Williams Syndrome and Down's Syndrome: Number in the infant and mature phenotype

Both WS and DS have been reported as having impairments in the number domain in adulthood, although the research conducted with WS has been hitherto sparse. This thesis presents the first work to characterise the precursors to number in atypical infants and to investigate whether the pattern displayed in the mature phenotype is already present in the starting state. Again, an attempt was made to use similar tasks with infants and adults. Both tests involved measuring numerosity discrimination. However, as mentioned in the chapter on infant number (Chapter 6), these tasks may not be measuring exactly the same cognitive processes. The infant task involves discrimination between novel and familiar numerosities which is indeed likely to tap number-relevant cognitive processes, but the adult task requires an understanding of the semantics of number. The cognitive process used in adulthood is more complex. However, the process used for the infant task is likely to be important and underlie the development of the adult competence.

8.3.1 Number in the infant phenotype

Sensitivity to changes in numerosity was measured in infants with WS, DS, and MA- and CA-controls. Given the number difficulties of both groups in adulthood, it was predicted that if initial states can be simply inferred from endstates, then both the WS and DS group should show impairment on this task. In line with my theoretical stance, the results did not support this oft-held assumption (e.g., Baron-Cohen, 1998; Leslie,

1992; Temple, 1997). The results indicated that the CA-matched, MA-matched and WS groups looked longer at the novel numerosity and were therefore extracting numerosity information from the displays. By contrast, the infants with DS looked equally long at both the novel and familiar displays and did not appear to extract numerosity information. It is important to note that this difference was not an artefact of familiarisation time, because looking time in both the WS and DS groups decreased by a similar amount across the familiarisation trials. The data are consistent with the claim that one aspect of the precursors to number, sensitivity to changes in small numerosities is present in infants with WS, but not in those with DS. This is striking given the adult data, to be presented below. As we shall see, the pattern of number ability found in infancy does not reflect that seen in adulthood.

8.3.2 Number in the mature phenotype

Numerosity discrimination in the mature phenotype was assessed using a task which was as similar as possible to the infant task, so that comparisons could be made across development. Across both the random and canonical dot experiments the MA-matched, CA-matched and DS groups exhibited the symbolic distance effect. For Arabic numerals, the effect was significant for the control groups and approaching significance for the DS group. However, the WS group did not exhibit a significant effect for any of the tasks, although the differences were in the expected direction. This suggests that they might represent number more weakly or in a different way, and be less affected by the semantics of numerosity, as represented by its analogue code. In these individuals, the balance between semantics and other factors may be different from that present in normal adults. Although the DS group did not demonstrate one of the fundamental biases for number in infancy, they had acquired a basic conception of numerosity by adulthood. On the other hand, the infants with WS appeared to exhibit this precursor for small numbers, but are likely to follow a different trajectory in subsequent learning, because they fail to display the same competency as the CA- and MA-matched adults with DS.

Data from a further study in the thesis support the idea that numerosity representations in WS are unusual. Participants underwent a battery of number tasks,

used with neuropsychological patients. Despite being matched on CA and MA, the DS group performed significantly better overall than the WS group, with the WS group exhibiting particular difficulties with ordering numerosities when they could not rely on rote strategies. This points to a possible problem with the underlying semantic representations of numerosity in the WS group.

It may be that the representations of numerosity of people with WS are weaker than in the other groups so that they experience greater interference when discriminating two numerosities, irrespective of the numerical distance between them. Such a problem with the representations would dilute the effect of distance because there would be interference throughout the system. Although somewhat increased for numerosities that are close together, this would be less marked than normal.

These individuals may also have a differently structured number system. While they are able to deal with number as individual lexical units, “one” or “1”, they may have an ill-defined or poorly structured system of numbers. Their phonological representation of the numbers may be clear, but they may have a weak conceptual system. This means that they may not fully understand the number system, in terms of the rules for manipulating numbers and how they are related to one another. This would have consequences for basic number processing and for using numbers in mathematical operations.

8.3.3 Summary

In summary, while infants with WS appear to be able to extract numerosity information from a visual display, adults with WS demonstrate impaired performance on a numerosity comparison task. Although they do show a trend in the correct direction, their lack of a significant distance effect suggests individuals with WS may be using a different strategy to deal with numerosity, which they continue to rely on, with less success, for tasks involving more complex numerical processing, such as those which demand an understanding of the semantics of number.

The opposite pattern of infant and adult performance obtains for the DS. In the DS group, it appears that numerosity understanding is delayed in infancy but that by adulthood, people with DS are using similar numerical processes to normal controls.

Again, these data highlight the importance of characterising the infant phenotype. If the adult profile had been used to infer the infant profile, the presence of different starting states and divergent developmental trajectories would not have been discovered.

8.4 Conclusions

The research in my thesis was conducted to investigate language and number understanding in the infant phenotype of Williams Syndrome and Down's Syndrome and to ascertain whether the pattern seen in infancy could have been simply derived from that present in the mature phenotype, as many theorists have assumed (e.g., Baron-Cohen, 1998, Leslie, 1992, Pinker, 1994, Temple, 1997). This is the first time that an empirical evaluation has been made of the oft-held (albeit implicit) assumption that the infant profile can be inferred from the adult profile. This assumption has important implications for the modularity continuity hypothesis. This states that the brain is organised into innate mental modules which are present at birth and which hold constant across the lifespan. Consequently, it implies that infant and adult profiles would be the same. In a developmental disorder, if a module were found to be impaired in adulthood, it would ex hypothesis also be impaired in infancy. In order to assess the assumptions of the modularity continuity hypothesis, the results from studies with atypically developing infants and controls were presented along with data from older children and adults. Although others have argued this theoretically (Bishop, 1997; Karmiloff-Smith, 1998), this thesis is the first to show empirically that the infant cognitive profile cannot be inferred simply from the mature phenotype in atypically developing groups. This casts doubt upon the modularity continuity hypothesis and points to the crucial importance of infancy research.

8.4.1 Theoretical implications

The results in my thesis emphasise that neurodevelopmental disorders should not be thought of in terms of static impaired and intact modules, present at birth. Instead, the data highlight that very different outcomes can arise from similar starting states and that different starting states can result in similar outcomes. This is illustrated by the vocabulary data in which both the DS and WS groups were delayed in infancy but the WS group exhibited an advantage in adulthood. It is likely that vocabulary acquisition followed a different trajectory in each group. Such differences in developmental trajectories in atypical groups mean that it is crucial to study the process of development and not merely to make assumptions using data from the endstate.

The results also have implications for typical development. Until recently, researchers inclined to posit the existence of innate principles in various cognitive domains, for example, in theory of mind or number, have tended to assume that these principles were generally similar to and operated in much the same manner as adult principles (Leslie, 1992; Wynn, 1992.).

Let us take theory of mind as an example of the implicit reasoning that calls upon the modularity continuity hypothesis. Leslie (1992, 1994) argues for the existence of a theory of mind module from the findings of research into Autism, another developmental disorder. He shows that older children and adults with Autism are poor at understanding false beliefs but that they are successful in understanding other “false” situations, such as out-of-date representations, like photographs. From this, he argues that there is a dedicated system for understanding mental states and that it is separate from understanding other representations. This would lead to an impaired theory of mind with other representations intact. In Autism, “very early biological damage may prevent the normal expression of this theory-of-mind module in the brain resulting in the core symptoms” of the disorder (p. 21). In terms of the modularity continuity hypothesis, this suggests that there is an inbuilt theory-of-mind system in the brain. The argument takes impairments which were found in older children and adults and uses these to posit the existence of an innate module. This would lead to an

assumption that the same impairments would be present in infancy. However, Leslie has shown only that the endstate, when all the crucial development has already taken place, is impaired. It could be that it is a different developmental trajectory which causes problems with theory of mind and not an impaired innate module, but this possibility is never discussed. The modularity continuity hypothesis is simply assumed.

New developments in the field of infant cognition have made it clear that innate principles are likely to be far more limited in scope than was first believed (Elman et al., 1996; Karmiloff-Smith, 1998; Karmiloff-Smith et al., submitted). My data highlight the complex nature of the link between genotype and phenotype, indicating that straightforward mappings from genes to behaviour are most unlikely to be viable. The data suggest that despite similar starting states, remarkable differences in the endstate can emerge. I believe that in order to explain the different developmental trajectories observed in the atypical groups examined in this thesis, a more dynamic approach to development is necessary. Such an approach is described below.

8.4.1.1. A dynamic theory of development – neuroconstructivism.

As mentioned above, the link between genotype and phenotype is likely to be complex, with both genes and environment playing a role in brain and cognitive development. In addition to this complex interaction between genes and the environment, development takes place against an ever-changing background. It is likely that the real process of development is far from that suggested by the adult neuropsychological literature, with pre-designated modules triggered by some environmental stimulus or maturational timetable. But it is also unlikely that the infant enters the world with no biases and simply waits for the environment to totally shape development. Instead, neuroconstructivism allows for the infant to contribute to the developmental process some pre-existing groups of neurons and processing skills, which are domain-general but domain-relevant and *become* domain-specific after processing relevant aspects of the environment. However, it does not suggest that these are pre-wired representations of higher cognitive processes or knowledge, such as the past tense rule for verbs. Such an approach is taken by a number of leading

developmentalists both in cognitive science and neuroscience (see Elman et al., 1996; Quartz & Sejnowsky, 1997; Karmiloff-Smith, 1998; Oliver et al., 2000).

What the infant does possess is a relatively plastic and adaptable brain which is endowed with several different neuro-architectures which are likely to be particularly suitable for the processing involved in higher cognitive functions. To clarify, there may be some aspects of neural circuitry which form a feedback mechanism, for example. This type of mechanism might lend itself to tasks involving serial processing, such as language. While it is likely that such architectures pre-exist, this does not mean they have been assigned a pre-existing function, in the sense of innate modules. What these architectures provide are particular modes of processing which may be more suited to one type of input over another but which are adaptable according to the developmental pathway the infant itself follows.

This control of development by the developing organism itself is another important tenet of this dynamic approach. By this view, the developing infant is an active participant in development, not a passive bystander waiting for development to happen to him or her. The infant contributes to the developmental process in a number of ways. In particular, this is achieved by the aspects of the environment on which he or she decides to focus and how that information is processed. Let us take Williams Syndrome as an example of how this might work.

Infants with Williams Syndrome focus the majority of their attention on faces at the expense of attention to objects (Mervis et al., 1999). This may cause subtle changes in the environment in which they find themselves for the following reason. The extra attention paid to the faces of adults around them may elicit more linguistic input than usual and this, in turn, might help to “tune” up those neural architectures which are suited to the processing of the serial input provided by language. This might be an indicator that large amounts of processing resources have been given over to language input. This could be at the expense of other types of input. Certainly, it seems that WS is less lateralised for language process than in the normal case (Neville et al., 1994). We do not yet know why WS infants pay so much attention to faces. It may be that from the outset the language-relevant networks in their brains are privileged and

looking at faces provides the language input sought by infants to make use of such networks. This verbal input would be well suited to the architecture and would further tune the networks. Or it may be that the face preference comes first.

Many networks would be available to the developing infant, but differences in the timing of gene expression, gene dosage, the timing of neural development or in the way in which neurons form connections with each other might cause some of these architectures to be more effective or more prevalent than others. This is where the neurodevelopmental disorder would have its first effect.

Consider the development of exemplar learning versus rule abstraction. It is clear how concentration on one type of process might have detrimental effects on others. The presence of neural structures that are particularly suited to linking individual inputs to individual outputs would create an environment in which individual exemplars can be represented. If an individual is able to represent individual exemplars and finds this a useful strategy then perhaps when development does not progress normally they will persist with this strategy and rely less and less on other forms of representation as development progresses. This could result in a bias towards exemplar-based representations at the expense of representations in which rules are extracted, prototypes are formed and individual exemplars are discarded. Over time, an already weaker system could become almost unusable, with many of the neural resources invested elsewhere. So, an individual with WS may form representations of individual exemplars of particular words but be less efficient at producing a prototype of that word. This proposition underlies the hypothesis that phonology in WS may be over-specified, at the expense of more abstract semantically-based representations of words. In terms of my data, such a strategy might explain early language delay because it is not the normal path to vocabulary acquisition. While infants with WS may link the spoken word to an object or event, they may fail to generalise this to different objects and events. Their relatively good vocabulary in adulthood could be due to an accumulation of processing resources dedicated to this domain.

The dynamic approach to development also allows for small differences at the outset of development leading to large effects in cognitive functioning later on. A small

change in the timing of gene expression or the migration of neurons can lead to a change in the type of neural circuitry available to the infant at a particular time and may cause the infant to be biased towards one type of input over others. Although certain neural architectures are likely to be suitable for different types of input, the brain is very plastic and if some circuitry is not working as well as others, those mechanisms that are functioning will process the input that would have been more suited to the disrupted mechanisms. This means that subtle deficits could be found in many areas of cognition in which inputs are being processed in a slightly different way. For example, take Specific Language Impairment. An adult with SLI might be able to compensate for his language difficulties using strategies developed over the course of development, borrowing from the more efficient areas of his brain. However, in subtle tests investigating, for example, the processing of rapidly changing auditory stimuli, the underlying deficits may become more apparent. This is because in order to do this type of rapid processing, the types of architectures which may have been disrupted are needed. Indeed, empirical work has suggested that this might be the case (see Tallal et al., 1996).

This neuroconstructivist approach does not claim that there is no specialisation. Rather, it is the process of development that causes such specialisation to emerge and become consolidated. The brain has neural pathways which may be suited to particular types of input and, with continued use and exposure to such input, become increasingly specialised. However, these architectures wait to be recruited by the developing infant and can be used either for something for which they are well-suited or can be co-opted to fulfil a role that is perhaps not ideally suited to them but which works for the most part.

The shaping of the brain by genes, the environment and the behaviour of the infant itself can lead to a subtle pattern of strengths and weaknesses in developmental disorders. In the following paragraphs, I will suggest how this theory might be applied to some neurodevelopmental disorders other than Williams Syndrome.

In Down Syndrome, brain development may not be as specialised as that seen in typical development. Instead of gradually specialising over development, individuals

with DS may continue to use several types of neural structures to tackle one type of input. This would mean that the particular constraints of particular systems which make them suited to language or number are not used effectively. In this case, systems which might not be directly relevant for language may be employed for language input to detrimental effect. This lack of specialisation could cause diffuse cognitive impairment because no particular input is preferred and therefore no particular mechanisms are fine-tuned. That said, visual processing is certainly superior to auditory processing in DS, so it is possible that the constraints and micro-circuitry suited to visual input are less affected than those suited to auditory input. Auditory input may be dealt with using slightly less suitable circuitry and in turn may be a less attractive stimulus for these individuals.

Possible explanations for the complex pattern of impairments in Autism could also be provided by this approach. Instead of considering the disorder in terms of an impaired module relating to say, meta-representation or theory of mind, it could be explored in terms of a more diffuse impairment in neural structures or processing which has more detrimental effects on some types of inputs than others. Impairments to the structures involved in one type of processing would have a graded effect on various aspects of cognition. It could have a great effect on social relations, because the resulting architecture would be less suited to these kinds of representation. By contrast, the effects may be weaker on vision, for example. The abilities which are highly developed in people with Autism may emerge because they rely on those forms of processing to which their brains have become particularly suited over the course of development. As an example, the enhanced perception of features may arise from a particular type of specialised processing in the autistic brain. However, such processing could be detrimental to other forms of cognition. The featural processing style of such individuals might lead to good performance on tasks such as the embedded figures test but could cause difficulties when trying to decouple the real features of an object, a banana for example, when it is being used in place of a telephone in pretend play.

The work in this thesis suggests that the neuroconstructivist approach to development is likely to be extremely useful for the study of neurodevelopmental disorders because

it allows for the complex interaction between the infant, its environment and biological constraints. This approach enables one to begin to unravel how small deviations from the norm early in development can have large effects later on and how similar endstates can be reached by many different means.

8.4.2 Research implications

My data suggest that the cognitive profiles in the mature phenotype are not necessarily a reflection of “set” patterns present at birth. Rather, one must look to the complex and ongoing interaction of multiple factors to determine precisely how these patterns come about. As a developmental cognitive psychologist, I believe that future research should concentrate on investigating these complex interactions by charting developmental trajectories from infancy onwards, rather than merely taking developmental snapshots.

In order to do this several methodological points should be considered. First, where possible, new research should be conducted longitudinally, so that subtle changes in the developmental trajectory can be detected both within an individual and across groups.

Second, the focus on atypical development is extremely important, because this can provide a window on the possible causes of differing developmental trajectories. For example, the comparison of DS and WS groups suggests that differences in the developmental trajectory are not due solely to general cognitive factors. Both groups were similarly impaired overall. The research in this thesis did not aim to examine the possible causes of a difference in the trajectory, and further work is necessary to examine them. One possibility is that the developmental trajectories in WS and DS may be influenced by subtle differences in processing biases or constraints used throughout development. For example, differences in the biological factors which influence the development of the brain, such as the formation of connections between neurons, may lead infants with WS and DS to attend to different aspects of a stimulus or process them differently (Karmiloff-Smith, 1998; Oliver, Johnson, Karmiloff-Smith and Pennington, 2000). These biases would then have an effect on further

development. The infants might pay special attention to the kinds of stimulus to which the way in which their brain developed is particularly suited. In WS, it could be that there are larger than normal areas of the brain given over to language which leads to good representations of single examples of words. However, the amount of space given over to language may mean that individuals are less able to extract rules and build a systematic representation of language.

Third, tests that are similar across wide age ranges should be used where possible. This was an aim of this thesis. This is important because one cannot argue that there are changes in cognitive profiles over development if one is measuring different aspects of a domain at each stage. So, for example, difficulties with number conservation in adults with WS should not be directly compared with differences in numerical discrimination in infants, because they tap different aspects of number. By contrast, the adult number discrimination task used in this thesis has more in common with the infant task. Although the tasks used tap different stages of number development and possibly different underlying processes, they are likely to be tapping abilities that are on the same developmental course in normal development. Discriminating between small quantities is likely to be important for the understanding of number meaning. The level at which one tests a particular ability is also important if taking a developmental approach. It is important, where possible, to use subtle tests which assess cognitive processes, rather than relying on a standard score. The latter only tells you the performance level of an individual, not how it was achieved. For example, research on face processing in WS has shown that individuals achieve normal scores on standardised tests, but the means by which these are achieved are very different from normal (Karmiloff-Smith, 1997).

Finally, the research has highlighted the importance of examining development from as early as possible. If one starts in middle childhood, one already misses important parts of the developmental trajectory. This is because the vast majority of brain and cognitive development occurs in the first few years of life and would have greatly slowed or ceased in older children and adults. On the other hand, if one starts to study developmental disorders in infancy and follows development, it should be possible to see if or where the developmental trajectories of different groups diverge. By

examining development closely, it may be possible to see how differences very early on change the way in which the infant interacts with the environment. For example, we know that young children with WS show a greater than normal interest in faces and language (Mervis et al., 1999). Over the course of development, this bias towards faces may mean that the infant receives more social contact and thus more language input. Cascading effects over development could lead to a relative strength in language over other types of cognition and this in itself may alter learning and the way in which other types of input are processed.

The research in my thesis was an attempt to follow a truly developmental approach. However, it was not able to adhere to all the principles suggested by the data. Given, that this thesis was prepared over a three-year period, a full longitudinal design with atypical groups was impractical. This should be attempted in the future. In addition, I was unable to see very young infants. Those with WS are often ill in the early months of life and so are untestable, and until recently, diagnosis was often not made until late toddlerhood or beyond. Now, we are beginning to recruit younger infants, diagnosed soon after birth, and I will be including them in my postdoctoral research.

Where possible, I have tried to keep tests as similar as possible across age groups. The present research has highlighted the shortcomings of various tests, both standardised and experimental. It can be extremely challenging to use standardised tests with populations on which they have not been normed. This is not only because the calculation of standard score is not possible, but also because test conditions can be inflexible. Nonetheless, one cannot dispose of standardised tests completely with clinical groups, but they should be administered with caution. It is also important to note that experimental studies are more sensitive and highlight strengths and deficits that are often missed by standardised tests. This sensitivity is crucial when testing atypically developing groups.

8.4.3 Further research

The following section will examine some methodological issues and suggests ways in which the research presented in this thesis could be furthered.

Infant studies

The infant studies presented in my thesis employed methods which have in the past been used with much younger typically developing infants. The studies in this thesis constitute the first use of preferential looking methods with children with Williams Syndrome and Down's Syndrome and were in the main successful. However, in future research improvements could be made.

First, parents of infants with DS remarked that their infants preferred photographs of real objects to line drawings. In the language study, the stimuli were photographs, but this was not the case in the word order or number discrimination studies. While there was no problem with engaging the attention of infants in the number study, there were problems in the word order study. New state of the art video technology will enable us to present not only photo quality images, but also moving stimuli. It is expected that dynamic stimuli will be particularly engaging and will make the actions taking place in the word order stimuli much more salient. This should mean that infants remain on task for longer and are more acutely aware of the aim of the "game" in which they are participating. This forms part of my current postdoctoral studies.

Second, as mentioned above, the infants in this study were much older than those normally tested using the preferential looking technique. While younger typically developing infants can be kept relatively still in the Fagan apparatus and are content to sit passively and watch the displays, older atypically developing infants are much more difficult to engage. In my studies I was careful to make the stimuli as attractive as possible and to engage the infant between trials, but there were aspects of the procedure which could be improved. The method of presentation in the Fagan box, where stimuli are manually inserted and removed, means there are breaks between trials which can demotivate the infant. This is especially true when she gets a brief glimpse of an adult but is prevented from a full-scale social interchange. Here again, a computerised set-up helps to surmount this problem. All the trials are already set up before the test begins, so data collection can go ahead with no breaks. The interval between trials can also be used to grab the infant's attention, because the computerised displays allow for the presentation of attractive moving fixation points.

Third, the nature and length of familiarisation trials should be addressed. In this thesis, two studies employed familiarisation: the word order and numerosity discrimination experiments. The familiarisation times reported for these studies with typically developing infants were increased for the experiments in this thesis to allow the infants with WS and DS more time to process the stimuli. Although no difference was found in the extent to which infants with WS and DS familiarised to the number stimuli, it might be argued that the DS group needed longer to familiarise to the stimuli. In order to guard against the possibility that infants do not receive a long enough familiarisation phase, it might be prudent to use infant-controlled familiarisation, rather than fixed familiarisation lengths. In order to do this, it is necessary to employ an automated procedure, which was not available at the time my studies were carried out. However, a computerised system could be used to keep track of infants' looking patterns and to curtail familiarisation when the infant reaches a pre-specified criterion. This would ensure that each infant had all the familiarisation time that she required. This could be particularly useful when testing atypically developing infants, because they may need longer to process the stimuli with which they are presented.

Adult studies

The research with older children and adults presented in this thesis was intended as a benchmark against which to compare the infant data. I characterised receptive vocabulary ability and two aspects of syntactic processing in WS, DS and control groups. As discussed in Chapter 5, the syntactic processing studies could be improved. An attempt was made to take an existing paradigm and make it more participant-friendly. However, this was not entirely successful. In future research, two aspects of the study should be considered. First, the memory load of the study should be reduced if possible. This should enable all the subjects to make complete responses which can then be analysed. Second, the studies highlighted a possible confound between sentence length and grammatical complexity. Stimuli in future experiments should be designed so that the effect of sentence length and memory load

on responses can be considered independently. To do this, stimuli should be made up of two types of sentence of the same length but differing complexity.

In addition to the language studies, I investigated numerosity comparison and basic number skills in older children and adults. This was the first investigation of its kind with the WS group, and therefore, as mentioned in Chapter 7, further, more detailed studies should be carried out.

8.4.4 Themes in the Williams Syndrome data and a future research programme

One particular theme has recurred in the data gathered from the Williams Syndrome group, both in language and number as well as in infancy and adulthood. This may be an indicator of the particular processing biases which give rise to the WS developmental trajectory. From my data and other data from our research group (see Karmiloff-Smith et al., submitted), it appears that individuals with WS place relatively less weight on semantic information compared to other sources of information.

In the context of this thesis, this problem with semantics is indicated by the lack of a robust distance effect when making number comparisons. The distance effect is said to reflect the presence of a semantic representation of number, in the form of numerosity. It might be that adults with WS do not have a clear understanding of the meaning of number, but are able to rely on the phonological system when counting by rote. This is highlighted by data from the number battery in which, when an understanding of the meaning of number was necessary, the performance of the WS group was poor.

The under-utilisation of semantics in WS may also be present in the language data. The literature review highlighted the unusual developmental trajectory of this group for lexical acquisition. It appears that they learn object labels (a phonological representation) before they can categorise objects (a semantic representation) (Mervis

et al., 1997). In this thesis, it was found that individuals with WS showed an advantage over DS in vocabulary tests in adulthood but that they retained some difficulty with syntax. Their over-reliance on phonology and under-reliance on semantics may cause problems when they are processing syntax. Phonological representations allow for the acquisition of single words but these are not sufficient for the development of syntax. Further cognitive processes are necessary for this. Semantics is likely to underlie one of these processes. In order to decode sentences, individuals must be able to integrate both lexical and syntactic information. In contrast to this pattern, it might be that the DS group is especially reliant on semantic information in language processing because of their difficulties with phonological processing. Data from studies of syntactic understanding in children with DS hints at this. As mentioned in Chapter 4, Fowler (1990) suggests that children with DS rely on semantics at the expense of syntax when decoding word order.

While this is an interesting emerging theme, it needs to be examined further. My future research program should be able to contribute to the investigation of this in early language development. Following on from my findings about vocabulary performance in atypically developing groups, I shall examine how infants with WS actually go about acquiring their lexicon. Research with children and adults, as mentioned in Chapters 4 and 5, has suggested that lexical development in WS follows an atypical trajectory. Given the results of this thesis, my future research will focus on the infant starting state and will use preferential looking methods to investigate the constraints in operation during vocabulary acquisition. By focusing on the very early stages of development, I hope to detect the point at which the WS developmental trajectory deviates from that seen in typical development. The preferential looking paradigm will enable me to subtly manipulate the constraints which could be used by an infant to acquire new words, and in this way I can attempt to characterise those which are important for infants with WS. It may be that infants with WS pay less attention to semantic cues, so their absence would have less of an effect on them than on typically developing infants. In contrast, a manipulation of the phonological cues available may cause them particular difficulty. Initially, I will investigate in infancy the role of the whole object, mutual exclusivity and taxonomic constraints in the acquisition of new words, building on previous research with adults (Stevens &

Karmiloff-Smith, 1997). However, in the future I will expand the work to examine the role of both phonological and semantic factors in lexical acquisition. To do this, infants will be taught new words in situations where the contribution of semantic and phonological cues is varied. This should enable me to pit one against the other.

8.4.5 Last word

In conclusion, I believe that my findings have important implications for cognitive science and cognitive neuroscience. The data contribute to both research and theoretical considerations and provide several exciting springboards for future investigations, especially in atypical infancy research.

Appendix A: Sentences for Imitation Task

Subcategorisation sentences

1st sentence is grammatical version, 2nd sentence is ungrammatical version

Neutral sentences

The girls in the park were eating apples and a huge packet of crisps.

The girl had beans on toast for supper.

Test sentences

I never listen to the radio.

I always listen the teacher.

I always laugh at him when he tells good jokes.

She always laughs him when he tells silly jokes.

Mary screamed at the dog which was running towards her.

Jenny screamed the spider which was crawling past her.

She arrived at the station just in time for her train.

He arrived the airport in time to meet his son.

I looked at five different books before I found the right one.

She looked six different coats before she found the right one.

I apologised to him because I felt so guilty.

She apologised me because she broke the vase.

When Laura tidied up she got rid her old clothes.

When Alice tidied up she got rid of lots of rubbish.

She stopped when the tour guide pointed to the lovely statue.

She stopped when the tourist pointed the ancient temple.

I don't like arguing, so I often try to agree with her.

Robert was convinced of the truth, so he could not agree me.

When I go on the train, I try not to speak to other passengers.

When she goes on the bus, she likes to speak other passengers.

Test sentences correct without preposition

I helped the girl to carry her shopping.

He helped the girl with her homework

I always hear my mother when she calls me

I always hear to my sister when she cries.

The teacher showed the chocolate cake to the girls.
The teacher showed to the cake to the boys.

The doctor advised her to eat less fat and more fruit.
The nurse advised to him to take much more exercise.

I hope I can forgive to the thief who stole my wallet.
I hope I can forgive the little boy who broke my vase.

After lunch I sometimes watch at television.
After dinner I sometimes watch football matches.

Mary was lost so she asked the man for directions
Bob was lost so he asked to the boy for directions

The last train had already left before Andrew reached the platform.
Uncle Jim had arrived before the girl reached to Auntie's.

The angry teacher could hear laughing before he entered to the room.
The nervous girl took a deep breath before she entered the exam room.

Word order sentences

Neutral sentences

We go to a football match every weekend.
Anna wore a very unusual costume to the fancy dress party.

Test sentences

He couldn't find the red pen anywhere.
She couldn't find blue the book anywhere.

When it is hot I always put on my hat and t-shirt.
When it is cold I put on hat my and gloves.

She has gin and tonic when she goes to the pub.
I have tea and cake when I go the to cafe.

I like to sit by the fire when it is chilly.
I like to walk the in park when it is sunny.

Bob was frightened of the big brown bear at the zoo.
Jane was frightened of the long snake green at the zoo.

I borrow four books from the library every month.
I borrow three books from library the every week.

I don't wear socks in summer because my feet get too hot.
I don't wear shorts in winter because legs my get too cold.

He met lots of exciting people at the office party.
I met lots of interesting people at birthday the party.

Gregory ate a huge coffee cake at his best friend Russell's party.
Alison baked a huge cake chocolate for her best friend Janet's birthday.

Steven was not looking because he was watching such an interesting match.
Adam was not listening because he was reading such interesting a book.

Short versions

Subcategorisation sentences

Neutral sentences

The girl had beans on toast for supper
The girls in the park were eating apples

Test sentences correct with preposition

I never listen to the radio.
I always listen the teacher

I apologised to him because I felt so guilty
She apologised me because she broke the vase

I laugh at him when he tells jokes.
She laughs him when he tells jokes

When Alice tidied up she got rid of rubbish
When Laura tidied up she got rid her clothes

I looked at five books before I found one.
She looked six coats before she found one.

She stopped when the guide pointed to a temple
She stopped when the tourist pointed a statue.

She arrived at the station in time for her train
He arrived the airport in time to meet his son.

Mary screamed at the dog which was running towards her
Jenny screamed the spider which was crawling past her.

I don't like arguing so I try to agree with her.
Robert was convinced of the truth so he could not agree me

When I go on the train I try not to speak to other people
When she goes on the bus she likes to speak other people.

Test sentences correct without preposition

I hope I can forgive the little boy.
I hope I can forgive to the thief

After dinner I watch football matches.
After lunch I watch at television

I helped the girl to carry her shopping.
He helped to the girl with her homework

I hear my mother when she calls me
I hear to my sister when she cries.

The teacher showed the cake to the girls.
The man showed to the cake to the boys.

The doctor advised her to eat much less fat.
The nurse advised to him to take more exercise.

The train had left before Andrew reached the platform.
Uncle had arrived before the girl reached to Auntie's

Mary was lost so she asked the man for directions
Bob was lost so he asked to the boy for directions

The girl took a deep breath before she entered the room.
The teacher could hear laughing before he entered to the room

Word order sentences

Neutral sentences

Anna wore a very unusual costume to the fancy dress party.
We go to a football match every weekend

Test sentences

When it is cold I put on hat my
When it is hot I always put on my t-shirt

He couldn't find the red pen anywhere
She couldn't find blue the book anywhere

I sit by the fire when it is chilly.
I walk the in park when it is sunny

She has beer when she goes to the pub
I have tea when I go the to café

I met lots of people at birthday the party
He met lots of people at the party

I borrow books from the library every month
I borrow books from library the every week.

I don't wear socks in summer because my feet get hot.
I don't wear shorts in winter because legs my get cold

Jane was frightened of the snake green at the zoo
Bob was frightened of the big bear at the zoo

Alison baked a huge cake chocolate for Janet's birthday
Gregory ate a huge coffee cake at Russell's party.

Steven was not looking because he was watching such an interesting match.
Adam was not listening because he was reading such interesting a book.

Appendix B: individual scores for language and cognition from the BSID II (Chapter 2)

Tables display individual percentage correct for language and cognitive items from the Bayley.

Williams Syndrome				Down Syndrome			
Name	Language	Cognition	Age*	Name	Language	Cognition	Age*
AG	77.78	78.05	1	FS	94.44	90.24	1
BP	94.44	93.9	1	FSch	83.33	81.71	1
JS	88.89	90.24	1	HS	88.89	90.24	1
SH	94.44	91.46	1	JB	77.78	91.46	1
TD	72.22	85.37	1	LA	94.44	96.34	1
ZR	88.89	87.8	1	SM	88.89	89.02	1
AW	75	96.34	2	SS	77.78	86.59	1
BP	79.17	95.12	2	TC	77.78	84.15	1
JC	95.83	93.9	2	AB	75	96.34	2
KC	70.83	95.12	2	AF	66.67	95.12	2
MS	83.33	96.34	2	CB	75	92.68	2
OD	100	95.12	2	EB	70.83	93.9	2
OT	83.33	91.46	2	EP	62.5	91.46	2
TD	66.67	97.56	2	EP	62.5	87.8	2
JC	100	93.26	3	GB	79.17	91.46	2
JG	96.97	95.51	3	SS	66.67	90.24	2
KC	84.85	88.17	3	AF	69.7	95.51	3
LMc	78.79	91.01	3	CB	69.7	91.01	3
MS	84.85	94.38	3	EB	57.58	93.26	3
OD	100	95.51	3	GW	84.85	94.38	3
RD	60.61	94.38	3	JK	63.64	95.51	3
				LP	84.85	94.38	3
				MC	81.82	98.88	3
				SR	66.67	95.51	3

Mental Age Matched				Chronological Age Matched			
Name	Language	Cognition	Age*	Name	Language	Cognition	Age*
AB	78.79	92.13	0	CB	81.4	98	1
AP	83.78	92.47	0	CM	81.4	96	1
CD	66.67	91.46	0	FC	83.72	98	1
DM	62.5	91.46	0	FI	86.05	94	1
HB	63.64	96.63	0	JT	88.37	96	1
HG	54.55	89.89	0	TM	79.07	95	1
HM	72.73	91.01	0	FH	79.17	93.4	2
HR	66.67	93.9	0	KE	89.58	96.23	2
IS	78.79	95.51	0	LB	91.67	95.28	2
KMH	87.5	98.78	0	NJ	87.5	90.57	2
LF	62.5	96.34	0	TH	89.58	92.45	2
LO	66.67	97.56	0	AH	94.23	95.61	3
MP	66.67	88.46	0	BH	94.23	100	3
NG	48.48	89.89	0	JA	92.31	94.74	3
SI	62.5	100	0	RC	90.38	95.61	3
SM	70.83	89.02	0	SS	92.31	93.86	3
TL	66.67	92.68	0	TM	84.62	89.47	3
TW	79.17	89.74	0				
VL	75	87.18	0				

*Age group codes

0 = MA matched group (varying ages)

1 = 24 months

2 = 30 months

3 = 36 months

Appendix C: individual mental age scores from the BAS (Chapter 3)

Tables display the mental age in months of individuals for seven subscales of the BAS and a mean mental age.

Williams Syndrome									
Name	Word definitions	Verbal similarities	Digit recall	Recall of designs	Pattern construction	Matrices	Quantitative reasoning	Mean	
RD	64	91	55	88	70	82	79	79	
RD	88	112	58	67	70	82	64	80.5	
CC	64	99	67	60	64	60	79	71	
CC	70	94	79	67	64	60	60	69.17	
JN	76	82	70	60	52	60	73	67.17	
JN	76	91	67	60	40	60	60	64.5	
AD	129	135	79	76	70	105	85	100	
AD	189	141	82	73	85	61	111	110	
GM	88	91	x	88	73	105	x	x	
GM	105	99	x	64	64	88	85	84.17	
LS	64	82	70	60	55	60	60	63.5	
LS	82	78	67	60	58	88	60	71	
TS	60	82	70	60	34	88	64	64.67	
TS	76	76	171	60	55	60	60	64.5	
NM	123	105	70	67	67	79	64	84.17	

x = individual did not complete this task

Down Syndrome									
Name	Word definitions	Verbal similarities	Digit recall	Recall of designs	Pattern construction	Matrices	Quantitative reasoning	Mean	
EB	60	70	70	99	129	60	79	82.83	
DT	76	82	58	64	70	60	60	68.67	
AB	60	70	34	73	67	70	64	67.33	
MR	60	60	30	60	61	60	64	60.83	
CM	60	91	61	76	73	60	85	74.17	
JG	60	60	x	60	34	60	64	56.33	
JP	60	91	58	67	111	76	79	80.67	
SH	60	94	48	73	117	94	67	84.17	
PH	60	60	30	60	64	67	60	61.83	

x = individual did not complete this task

Appendix D: individual scores for production and comprehension from the MacArthur CDI (Chapter 4, Experiment One)

The tables display the percentage of items produced and comprehended by each infant, based on the MacArthur CDI.

Williams Syndrome				Down Syndrome			
Name	Production	Comprehension	Age*	Name	Production	Comprehension	Age*
BP	8.84	51.01	1	FSc	0.78	17.93	1
SH	6.06	7.32	1	JB	1.01	33.33	1
TD	3.79	21.72	1	LA	15.65	26.26	1
ZR	1.26	4.79	1	SS	0.5	43.4	1
AW	7.83	34.85	2	TC	5.3	31.56	1
JC	34.34	49.24	2	CB	5.56	37.12	2
KC	11.62	65.4	2	EB	0	21.21	2
MS	19.44	20.7	2	EP	2.27	47.73	2
OD	62.62	76.01	2	GB	3.79	47.47	2
TD	1.26	30.05	2	LP	8.58	79.54	2
JG	61.62	85.1	3	SM	1.26	19.44	2
JS	1.77	24.24	3	SS	9.09	55.3	2
KC	42.21	76.52	3	CB	16.91	65.15	3
LM	9.6	34.09	3				
MS	37.63	50	3				
OD	82.83	90.4	3				

*Age group codes

0 = MA matched group (varying ages)

1 = 24 months

2 = 30 months

3 = 36 months

Mental Age Matched				Chronological Age Matched			
Name	Production	Comprehension	Age*	Name	Production	Comprehension	Age*
AB	11.62	56.06	0	CB	56.56	89.39	1
AP	49.74	87.63	0	FC	83	87.9	1
CD	3.03	34.84	0	JT	65.4	80.6	1
DMM	1.26	21.21	0	KE	92.9	96.2	2
HG	0	7.83	0	LB	86.36	94.69	2
HR	0.25	7.32	0	NJ	35.86	96.46	2
IS	5.56	53.03	0	AH	96.71	96.97	3
LF	4.55	34.85	0	BH	96.21	98.48	3
LO	7.32	50	0	JA	100	100	3
NG	1	23.74	0	SS	100	100	3
OJ	0	8.08	0	TM	58.33	74.75	3
PN	0	6.31	0				
SI	5.56	41.16	0				
TL	0	42.17	0				
TW	8.08	6.51	0				
VL	4.04	15.15	0				

*Age group codes

0 = MA matched group (varying ages)

1 = 24 months

2 = 30 months

3 = 36 months

Appendix E: individual scores for sensitivity to subcategory constraint violations (Chapter 5, Experiment Two)

Tables display number of responses given in each of four categories for each group.

Williams Syndrome						
Grammatical Subcategorisation				Ungrammatical Subcategorisation		
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct	Ungrammatical change to correct
GM	13	2	2	7	12	0
JN	8	3	6	2	10	1
NM	19	0	0	19	0	0
TS	3	10	0	13	0	2
CC	17	1	1	4	14	0
AD	17	1	1	0	18	1
LS	14	1	2	9	6	1
RD	11	2	6	4	10	2
Down Syndrome						
Grammatical Subcategorisation				Ungrammatical Subcategorisation		
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct	Ungrammatical change to correct
CM	13	1	3	2	12	1
DT	10	0	9	1	8	0
EB	8	1	9	2	8	1
SH	13	4	2	1	14	1
JP	7	6	6	1	13	2

Chronological Age Matched									
Grammatical Subcategorisation				Ungrammatical Subcategorisation					
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical change to correct	Other
CB	19	0	0	19	0	0	0	0	0
ZS	19	0	0	2	17	0	0	0	0
MAGS	19	0	0	13	5	1	0	0	0
RP	19	0	0	15	3	1	0	0	0
CA8	19	0	0	2	17	0	0	0	0
RM	19	0	0	18	1	0	0	0	0
GP	19	0	0	18	1	0	0	0	0
CL	19	0	0	19	0	0	0	0	0
Mental Age Matched									
Grammatical Subcategorisation				Ungrammatical Subcategorisation					
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical change to correct	Other
S9	12	0	0	4	10	1	4	1	4
S2	18	0	1	11	8	0	0	0	0
JR	19	0	0	17	2	0	0	0	0
S10	18	0	0	10	8	1	0	1	0
JAMA	14	0	2	3	11	0	3	0	3
MA3	19	0	0	8	8	1	2	1	2
S4	19	0	0	14	5	0	0	0	0

Appendix F: individual scores for sensitivity to word order violations (Chapter 5, Experiment Two)

Tables display number of responses given in each of four categories for each group.

Williams Syndrome						
Grammatical Word order				Ungrammatical Word order		
	Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct
GM	x	x	x	x	x	x
JN	x	x	x	x	x	x
NM	10	0	0	0	9	0
TS	5	5	0	0	0	1
CC	9	0	0	1	0	8
AD	8	2	0	0	10	2
LS	9	0	0	0	0	0
RD	5	1	0	1	0	0
Down Syndrome						
Grammatical Word order				Ungrammatical Word order		
	Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Verbatim	Grammatical change to correct
CM	8	0	1	0	0	9
DT	4	0	0	6	8	2
EB	5	0	0	5	0	2
SH	5	3	1	1	1	8
JP	6	3	1	0	0	8

x = individual did not complete this task

Chronological Age Matched

Grammatical Word order					
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical Word order	
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical Word order	Other
CB	10	0	0	10	0
ZS	10	0	0	5	0
MAGS	9	0	0	6	3
RP	10	0	0	10	0
CA8	10	0	0	0	1
RM	10	0	0	10	0
GP	10	0	0	10	0
CL	10	0	0	10	0

Mental Age Matched

Grammatical Word order					
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical Word order	
Verbatim	Grammatical change to correct	Ungrammatical change to correct	Other	Ungrammatical Word order	Other
S9	6	0	2	2	4
S2	10	0	0	5	1
JR	9	0	1	5	0
S10	8	0	1	4	0
JAMA	10	0	0	0	1
MA3	10	0	0	4	0
S4	9	0	0	6	1

Appendix G: individual mean reaction times for number comparison tasks (Chapter 7, Experiments One, Two and Three)

Tables display the mean reaction times in milliseconds for close and far pairs of dot displays or digits for each group.

Williams Syndrome						
Dot comparison canonical		Dot comparison random		Arabic numeral comparison		
Name	Close pairs	Far pairs	Close pairs	Far pairs	Close pairs	Far pairs
AD	632.3571	555.5556	676.9	550.6667	1480.278	910.75
CC	875.8333	688.125	786.0556	725.6667	1232.857	1287.333
GM	1108.363	1533.677	4810.222	1983.333	950.25	795.7857
JN	3174.889	2964.944	1334.75	1359.083	2105.556	2170.857
LS	1305.556	1216.222	1305.556	1216.222	1349.111	1697.167
NM	914	818.7222	914	818.7222	1040.722	1272.944
RD	2141.944	978.2222	1256.056	889.6667	1458.278	910.75
TS	3036.688	2606.563	1207.444	907.4286	6184.778	5634.889
Down Syndrome						
Dot comparison canonical		Dot comparison random		Arabic numeral comparison		
Name	Close pairs	Far pairs	Close pairs	Far pairs	Close pairs	Far pairs
CM	2302.944	1971.389	4799	2848.444	2487.438	2544.944
DT	3528.222	3659.611	4628.167	3861.278	4818.722	3630.278
EB	1223.056	704.7222	1253	806.5	1276.167	973.6111
AB	1939.833	1497.313	2472.377	2070.48	4631.444	4097.889
JG	X	x	x	x	5954	3371.563
JO	799.5241	704.1118	1420.438	1184.938	1457.143	1284.222
MR	1169.222	916.3333	1135.056	989.3889	1408.188	1304.833
SH	2055.861	1813.014	2845.556	1832.375	2369.722	2135.778

x = individual did not complete this task

The table shows that on several occasions an inverse distance effect was present for the WS group with displays close in numerosity exhibiting longer reaction times than those with numerosities which were far apart. This happened on only one occasion in the DS group.

Mental Age Matched						
Dot comparison canonical			Dot comparison random		Arabic numeral comparison	
Name	Close pairs	Far pairs	Close pairs	Far pairs	Close pairs	Far pairs
MA1	905.8333	775.4444	822.2778	661.2778	846.9444	826
MA2	1262.278	1063.333	1354.5	1025.556	1353.722	1220.778
MA3	971.0556	808.7222	1036.556	761.8889	1116.389	1135.333
MA5	577.8889	481	659.7778	526.7222	488.8889	476.2222
MA6	2144.444	1774.278	2315.278	2432.667	2615.833	2217.667
MA7	1631.722	1306.167	3632.278	1239.778	1854.667	1474.611
MA9	1255	1191.5	2255.778	2027.889	1841.056	1532
MA10	1927.444	1228.5	1283.778	1186.167	1845.5	1693.722
Chronological Age Matched						
Dot comparison canonical			Dot comparison random		Arabic numeral comparison	
Name	Close pairs	Far pairs	Close pairs	Far pairs	Close pairs	Far pairs
CB	440	402.875	412.625	382.4286	584.125	481
CL	526.0556	469.7778	441.5714	418.5556	511.2778	454.5
RP	809.4375	798.9444	888.0556	801.5556	804.8889	700.5556
RM	587.625	475.5714	587.625	475.5714	466.5556	421.8889
CA4	1154.357	1007.278	1135.75	960.4444	1449.75	1381
CA8	974.8125	801.3333	1058.278	832.9375	1033.056	846.4375
ZS	783.4444	630.1667	673.4375	597.7222	543.0714	499.125
GP	556.8403	472.6746	514.5982	447.0635	488.9167	438.1944

Appendix H: individual accuracy scores for number comparison tasks (Chapter 7, Experiments One, Two and Three)

Tables display the mean percentage of trials correct in comparisons of dot displays or digits for individuals in each group. This is shown for close pairs, far pairs and for the total number of items.

Williams Syndrome									
Name	Dot comparison canonical			Dot comparison random			Arabic numeral comparison		
	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total
AD	77.78	58.33	68.06	100	100	100	63.89	94.44	79.17
CC	77.78	58.33	68.06	72.22	94.44	83.33	63.89	66.67	65.28
GM	86.11	100	93.06	72.22	97.22	84.72	77.78	88.89	83.33
JN	88.89	100	94.44	94.44	100	97.22	77.78	94.44	86.11
LS	83.33	91.67	87.5	88.89	100	94.44	77.78	86.11	81.94
NM	75	100	87.5	66.67	88.89	77.78	52.78	63.89	58.33
RD	86.11	100	93.06	97.22	100	98.61	75	86.11	80.56
TS	94.44	100	97.22	86.11	100	93.06	69.44	91.67	80.56
Down Syndrome									
Name	Dot comparison canonical			Dot comparison random			Arabic numeral comparison		
	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total
CM	94.44	100	97.22	97.22	100	98.61	97.22	100	98.61
DT	44.44	55.56	50	63.89	80.56	72.22	38.89	55.56	47.22
EB	100	100	100	100	100	100	97.22	100	98.61
HP	97.22	100	98.61	75	91.67	83.33	77.78	97.22	87.5
JG	x	x	x	x	x	x	66.67	72.22	69.44
JO	100	100	100	100	100	100	100	100	100
MR	97.22	100	98.61	97.22	100	98.61	97.22	100	98.61
SH	100	100	100	100	100	100	100	94.44	97.22

x = individual did not complete this task

Mental Age Matched

Name	Dot comparison canonical			Dot comparison random			Arabic numeral comparison		
	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total
MA1	100	100	100	100	100	100	100	100	100
MA2	97.22	100	98.61	91.67	100	95.83	94.44	91.67	93.06
MA3	100	100	100	100	100	100	91.67	97.22	94.44
MA5	97.22	97.22	97.22	100	100	100	88.89	100	94.44
MA6	77.78	94.44	86.11	97.22	100	98.61	97.22	100	98.61
MA7	100	100	100	91.67	100	95.83	94.44	100	97.22
MA9	88.89	100	94.44	94.44	97.22	95.83	80.56	97.22	88.89
MA10	100	100	100	100	100	100	94.44	91.67	93.06

Chronological Age Matched

Name	Dot comparison canonical			Dot comparison random			Arabic numeral comparison		
	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total	Close pairs	Far pairs	Total
CB	100	100	100	100	100	100	100	100	100
CL	100	100	100	100	100	100	100	100	100
RP	100	100	100	100	100	100	97.22	100	98.61
RM	94.44	100	97.22	100	100	100	91.67	100	95.83
CA4	100	100	100	97.22	100	98.61	94.44	100	97.22
CA8	94.44	100	97.22	97.22	100	98.61	88.89	100	94.44
ZS	100	97.22	98.61	97.22	100	98.61	97.22	100	98.61
GP	94.44	100	97.22	100	100	100	100	100	100

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